Developing a psychoeducational intervention to support patients undergoing screening and/or predictive genetic testing for inherited cardiac conditions (the PISICC study)

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by

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Abstract

Background: Inherited cardiac conditions (ICCs) encompass two disease entities: Inherited cardiomyopathies and Inherited arrhythmic syndromes. The prevalence of ICCs is estimated at 3% of the general population and therefore causes significant morbidity worldwide and is a leading cause of sudden death in the young. Relatives of an affected person generally have a 50% chance of inheriting the condition and are therefore recommended to undergo cardiac screening and/or predictive genetic testing (PGT). Patients often find undergoing these tests stressful and are worried not just for their own health but also worry about the impact of the result on their family. There is a high rate of drop-out from follow up of family members who are diagnosed with an ICC and/or are found to be genetic carriers despite carrying a risk for sudden cardiac death. Psychoeducational interventions in ICCs are scarce and none have been developed specifically to support at-risk family members.

Aim: The overall aim of this study was to develop a psychoeducational intervention to support at-risk family members undergoing cardiac screening and/or PGT.

Methods: The studies conducted in this research were based on the Development Stage of the Medical Research Council framework for developing complex interventions and guided by Self Determination theory (SDT). The first phase of the research was a mixed methods systematic review which provided the evidence base for the experiences of adult patients at risk for ICCs undergoing screening and/or PGT. Building on this knowledge, in the second phase of the research, a qualitative study of 29 adult patients who had screening and/or PGT explored the preferences and priorities for a psychoeducational intervention. Themes were generated through Framework analysis with the application of SDT as a conceptual framework. In the

third and final phase of the research, the synthesis of the findings of the systematic review and qualitative study alongside patient and public involvement activities informed the model for the psychoeducational intervention.

Findings: The systematic review detailed the experiences of family members at-risk for an ICC, including the psychosocial impact of this process, and identified areas requiring further support and development in the care pathway. The qualitative study generated four main themes wherein the theme, Impact of the proband's story, is the reference point to a family's journey into cardiac screening and/or PGT and the themes, Leveraged autonomy, Harnessing competence and Relatedness in the social context of ICCs, all correspond to the basic psychological needs fulfilment within SDT to promote optimal adjustment and coping. The synthesis of the findings from the systematic review and qualitative study revealed the need for early assessment of psychological needs and a psychoeducational intervention focused on patients who receive a diagnosis or likely diagnosis and/or are genetic carriers for an ICC. Furthermore, support is needed for decision-making regarding lifestyle, management, and communication to other family members. The refined intervention model following the PPI activity is comprised of components that address the basic psychological needs of competence, autonomy, and relatedness with recommendations for timing, delivery, and outcome measures.

Conclusion: This research is a novel application of Self Determination theory to enable the development of an evidence-based psychoeducational intervention to support patients who have undergone cardiac screening and/or PGT with a new diagnosis or carrier status for an ICC. In addition, this study has identified multiple targets for service improvement along the ICC care pathway for at-risk family members.

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Abbreviations

ACMG	American College of Medical Genetics
AMED	Allied and Complementary Medicine Database
ARVC	Arrhythmogenic Right Ventricular Cardiomyopathy
BDI	Beck's Depression Inventory
BGCSES	Bergen Genetic Counselling Self-Efficacy Scale
BHF	British Heart Foundation
BrS	Brugada Syndrome
CAQ	Cardiac Anxiety Questionnaire
CAQDAS	Computer-Assisted Quality Data Analysis Software
CBM	Cognitive Behavioural Model
CBT	Cognitive Behavioural Theory
CGN	Cardiac Genetics Nurse
CINAHL	Cumulative Index of Nursing Allied Health Literature
cMRI	cardiac Magnetic Resonance Imaging
CPVT	Catecholaminergic Polymorphic Ventricular Tachycardia
CRD	Centre for Reviews and Dissemination
CRY	Cardiac Risk in the Young
CST	Cognitive Social Theory
DCM	Dilated Cardiomyopathy
ECG	Electrocardiogram
EP	Early Repolarisation
ESC	European Society for Cardiology
EXPLORER-HCM	Mavacamten for treatment of symptomatic obstructive hypertrophic cardiomyopathy
FH	Familial Hypercholesterolaemia
GC	Genetic Counsellor
GHQ	General Health Questionnaire
HADS	Hospital Anxiety & Depression Scale
HBM	Health Belief Model
HBOC	Hereditary Breast or Ovarian Cancer
HCCQ	Health Care Climate Questionnaire
HCM	Hypertrophic Cardiomyopathy
HD	Huntington Disease
HFA	Heart-Focused Anxiety
HLoC	Health Locus of Control
HR-QoL	Health Related Quality of Life
ICC	Inherited Cardiac Conditions
ICD	Implantable Cardioverter Defibrillator
IES	Impact of Events Scale
IPQ	Illness Perception Questionnaire

IRT	Interactionist Role Theory
IUT	Illness Uncertainty Theory
JLA PSP	James Lind Alliance Priority Setting Partnership
KCL	King's College London
KHP	King's Health Partners
KHP-PPIICC	King's Health Partners' Patient & Public Involvement for Inherited Cardiac Conditions
LQTS	Long QT Syndrome
LV	Left Ventricular
LVH	Left Ventricular Hypertrophy
LVNC	Left Ventricular Non-compaction Cardiomyopathy
LVOTO	Left Ventricular Outflow Tract Obstruction
MBSR	Mindfulness-Based Stress Reduction
MDT	Multi-disciplinary Team
MFDG	Multi-Family Discussion Group
MRC	Medical Research Council
Na+	Sodium
NGT	Nominal Group Technique
NHS	National Health Service
PCASS	Perceived Choice and Awareness of Self Scale
PCCD	Progressive Cardiac Conduction Disease
PCS	Perceived Competence Scale
PGD	Pre-implantation Genetic Diagnosis
PGT	Predictive Genetic Testing
PISICC	Psychoeducational Intervention Supporting Patients with Inherited Cardiac Conditions
PPI	Patient and Public Involvement
PRISMA	Preferred Reporting Items for Systematic Reviews and Meta-Analyses
QoL	Quality of Life
RCM	Restrictive Cardiomyopathy
RCT	Randomised Control Trial
REC	Research Ethics Committee
REMGC	Reciprocal Engagement Model of Genetic Counselling
SADS	Sudden Arrhythmic Death Syndrome
SADSUK	Sudden Arrhythmic Death Syndrome UK
SCD	Sudden Cardiac Death
SDT	Self-Determination Theory
SET	Self-Efficacy Theory
SGCS	Satisfaction with Genetic Counselling Scale
SIDS	Sudden Infant Death Syndrome
SQTS	Short QT Syndrome
TMSI	Threatening Medical Situations Inventory
TTM	Transtheoretical Model of behaviour change
UCM	Unclassified Cardiomyopathies

VF	Ventricular Fibrillation
VT	Ventricular Tachycardia
VUS	Variants of Unknown clinical Significance
WES	Whole Exome Sequencing
WGS	Whole Genome Sequencing

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Chapter 1: Introduction

1.1 Chapter overview

This chapter describes the background to the research, from its inception to the justification for the sequence of studies conducted. The current scientific knowledge regarding inherited cardiac conditions (ICCs) is discussed, followed by the impact of the condition on the health service, and on patients and their families. This is followed by an overview of various psychoeducational interventions in cardiovascular and genetic healthcare, and in ICCs with a justification for developing one specifically for family members at-risk for ICCs. The final section summarises the structure of the thesis.

1.2 Background and motivation for a research project

In 2008, my role as a cardiac genetics nurse (CGN) was part of a novel service development initiative of the British Heart Foundation (BHF). In the ensuing years, our ICC clinic grew, and I became an established practitioner. It was at this point where the journey of this thesis began. We were seeing more index cases (probands); however, we did not observe the expected increase in uptake of screening and/or genetic testing of their at-risk family members. We resolved this by improving access to the clinic through a self-referral system, but soon it became apparent that at-risk family members had their unique needs, and many were not attending follow ups or were non-adherent to the health advice given. In this group, their health education and psychological support needs seemed greater than their physical/medical needs because most were asymptomatic or had minor symptoms. Therefore, in these aspects, the demands on the nurses were far greater than on the doctors and we needed a more robust way to address these issues as the number of patients increased.

It was also at this time that I became more involved with patient support groups such as Cardiomyopathy UK, Cardiac Risk in the Young (CRY) and Sudden Arrhythmic Death Syndrome UK (SADS UK). My role in these groups was mainly to provide clinical expertise but I also listened to their stories-their tragic accounts of grave illness, sudden deaths in young people, the ongoing challenges of coping with a chronic genetic illness; and the fear of dying suddenly. I also sought their views in shaping our growing ICC service using a formal patient and public involvement (PPI) process. Whilst they fed back that they were satisfied with the service, it was felt that there was a huge gap in psychological support which hindered patients' coping, but the specifics of what this was and who provided it was unclear. It was this lack of clarity and the timeliness for the need of an evidenced-based intervention to support ICC patients that led me to propose the series of studies detailed in this thesis which comprises the Psychoeducational Intervention Supporting Patients with Inherited Cardiac Conditions (PISICC) project.

In planning and conducting the academic research recorded in this thesis, I have adhered to the highest standards of scientific methods and combined this with a pragmatic philosophical stance. I believe that this provides the epistemological justification in constructing a workable solution by combining multiple sources of knowledge to create an in depth understanding of the needs of ICC patients (Nowell, 2015). Pragmatism also lends itself to the realities in the National Health Service (NHS) where implementation of the outputs of this research is likely to require cooperative action from health professions and patients. It is my belief that ontologically, an external reality exists, but that this is interpreted uniquely by each individual based on their sociocultural backgrounds and other factors. However, as a society, we can identify commonalities and develop shared beliefs.

Aside from the patients themselves as a source of knowledge to fulfil the objectives of this research, the views of other patients, families and health professionals are also incorporated through the King's Health Partners' Patient & Public Involvement for Inherited Cardiac Conditions (KHP-PPIICC) group. This group was formed using established guidelines (INVOLVE, 2012) and to inform, oversee and maintain the relevance of the research.

1.3 Inherited cardiac conditions

The prevalence of ICCs is significant with a conservative estimate of 340,000 affected in the UK population (Burton et al., 2009). The combined prevalence of ICCs is estimated at 3% of the general population and therefore causes significant morbidity worldwide and is a leading cause of sudden death in the young (Girolami et al., 2018).

Inherited cardiac conditions encompass two disease entities: Inherited cardiomyopathies and Inherited primary arrhythmic syndromes. Inherited cardiomyopathies are a group of conditions affecting the heart muscle with no associated coronary artery disease and are further subdivided to include Hypertrophic Cardiomyopathy (HCM), Dilated Cardiomyopathy (DCM), Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), Restrictive Cardiomyopathy (RCM) and there is a category for Unclassified Cardiomyopathies (UCM) (Elliott et al., 2008). Inherited primary arrhythmic syndromes occur in a structurally normal heart with ion channel defects. These include Long QT Syndrome (LQTS), Short QT Syndrome (SQTS), Brugada Syndrome (BrS) and Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT), Unexplained cardiac arrest or Idiopathic Ventricular Fibrillation (VF) and Unexplained Sudden Cardiac Death (SCD) (Ackerman et al., 2011). Early repolarisation (ER) and Progressive cardiac conduction disease (PCCD) are also

described as primary inherited arrhythmias but can be a feature of the some of the ICCs mentioned such as Brugada (PCCD) or mechanisms of inheritance are not clearly defined (ER) (Basso et al., 2017).

Inherited aortopathies are also considered a subgroup of conditions under ICCs due to their cardiovascular features as well as their dermatological, joint and eye manifestations. They encompass pathologies of the aorta including aneurysmal dilatation and rupture. Examples include Marfan Syndrome, Loeys-Dietz Syndrome, Turner Syndrome, Bicuspid Aortic Valve, Vascular Ehlers-Danlos Syndrome and Familial Thoracic Aortic Aneurysm & Dissection (Fletcher et al., 2020). This PhD project is reflective of the cases referred to the specialist clinics in the centres where the research was situated, therefore the focus will be on inherited cardiomyopathies and arrhythmias.

1.3.1 Clinical features and diagnosis of inherited cardiac conditions

1.3.1.1 Inherited cardiomyopathies

1.3.1.1.1 Hypertrophic cardiomyopathy

Hypertrophic cardiomyopathy is characterised by left ventricular hypertrophy (LVH) without ventricular dilatation, where all cardiac or systemic causes of the extent of the LVH has been excluded. A diagnosis is made through echocardiography or cardiac magnetic resonance imaging (cMRI) when the left ventricular (LV) maximal wall thickness exceeds 15 mm. The threshold for diagnosis is lower for first degree relatives undergoing cardiac screening at 13-14 mm.

The distribution of LVH in HCM is usually asymmetric and about 70% of patients will develop LV outflow tract obstruction (LVOTO) which is an important cause of morbidity in HCM. Depending on the degree of LVOTO, patients may experience shortness of breath and blackouts. Around 50% of patients will experience disease

progression with a subset of 1% experiencing end-stage HCM with associated heart failure (Maron et al., 2006).

1.3.1.1.2 Dilated cardiomyopathy

The features of DCM include ventricular dilation and global dysfunction with an ejection fraction <50% (normal 55-70%) found on cardiac imaging without any associated coronary or systemic disease. This is a progressive condition which terminates with worsening heart failure, atrial and ventricular arrhythmias, stroke and/or sudden death (Cecchi et al., 2012).

1.3.1.1.3 Arrhythmogenic right ventricular cardiomyopathy

Arrhythmogenic right ventricular cardiomyopathy is characterised by infiltration of the myocardium with fat and fibrosis. Ventricular arrhythmias are a hallmark of ARVC and in later stages, ventricular systolic dysfunction is common (Marcus et al., 2010). The right ventricular form is more common, but it is now recognised that ARVC also affects the left side, hence, over the course of the research, the collective term for this condition has changed to Arrhythmogenic Cardiomyopathy (Corrado and Basso, 2021). However, for the purposes of this thesis, ARVC will be used as this is the term known to the patients at the time of their screening and/or genetic testing.

1.3.1.1.4 Restrictive cardiomyopathy

The main feature of RCM is a restrictive LV physiology. Cardiac imaging usually demonstrates reduced systolic and diastolic volumes, significant dilatation of the atria and flow impairment although LV wall thickness and systolic function are usually normal. Severe functional limitation with a poor prognosis is expected in RCM due to the extreme diastolic dysfunction resulting in reduced filling and resulting low cardiac

output. Restricted cardiomyopathy may also feature in end-stage HCM (Cecchi et al., 2012).

1.3.1.1.5 Unclassified cardiomyopathies

The UC are grouped as such because they are not considered disease entities on their own but rather a feature of the main cardiomyopathies. This includes left ventricular non-compaction cardiomyopathy (LVNC) where prominent LV trabeculae, LV dilation and dysfunction are present; and Takotsobu cardiomyopathy which appears as LV apical ballooning and features temporary systolic dysfunction in the absence of coronary disease (Elliott et al., 2008).

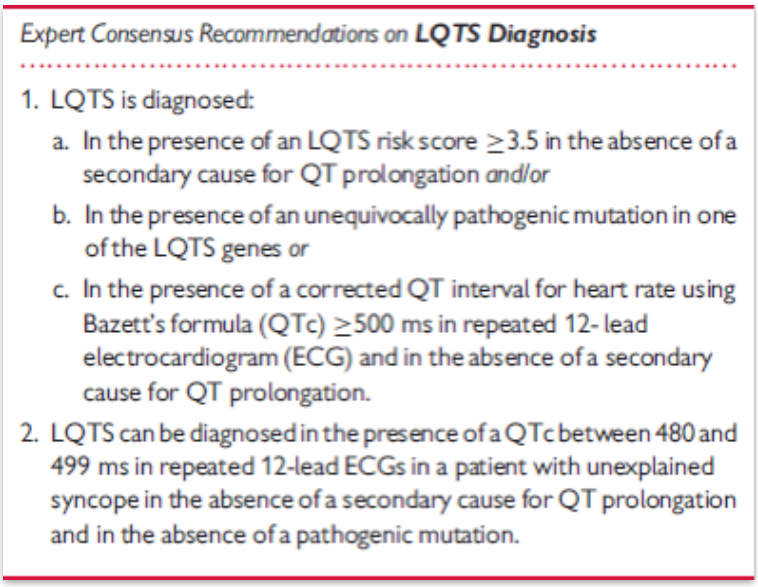
1.3.1.2 Inherited primary arrhythmia syndromes

1.3.1.2.1 Long QT syndrome and Short QT syndrome

In LQTS, there is a dysfunction in the ion channels which affects the cardiac action potential (Bohnen et al., 2017). Patients with LQTS experience arrhythmic events usually arising from a ventricular arrhythmia called torsades des pointes, which, depending on duration and severity, can lead to syncope, cardiac arrest and may progress to VF and sudden death.

Establishing a diagnosis of LQTS includes measurement of QT interval corrected for heart rate (QTc) on the electrocardiogram (ECG) based on Bazett's formula. Other causes of a prolonged QT must be excluded such as electrolyte imbalances, acquired conditions and QT prolonging medications (Priori et al., 2013). The diagnostic criteria are illustrated in Figure 1.

Figure 1 Diagnostic criteria for Long QT syndrome (Priori et al., 2013)



Expert Consensus Recommendations on ***LQTS Diagnosis***

1. LQTS is diagnosed:
   a. In the presence of an LQTS risk score ≥3.5 in the absence of a secondary cause for QT prolongation *and/or*
   b. In the presence of an unequivocally pathogenic mutation in one of the LQTS genes *or*
   c. In the presence of a corrected QT interval for heart rate using Bazett's formula (QTc) ≥500 ms in repeated 12- lead electrocardiogram (ECG) and in the absence of a secondary cause for QT prolongation.
2. LQTS can be diagnosed in the presence of a QTc between 480 and 499 ms in repeated 12-lead ECGs in a patient with unexplained syncope in the absence of a secondary cause for QT prolongation and in the absence of a pathogenic mutation.

Abbreviations: LQTS-Long QT Syndrome, ECG-electrocardiogram

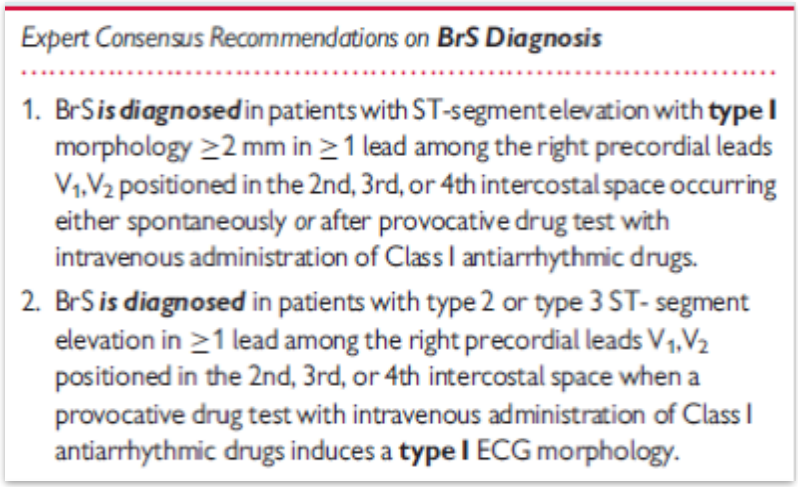
Individuals with SQTS may be asymptomatic or experience dizziness, palpitations. Atrial fibrillation or SCD (El-Battrawy et al., 2018). It is a rare condition and the diagnosis is made in the presence of a QTc ≤330 ms or a QTc <360 ms and the presence of one or more of these factors: the presence of a pathogenic genetic variant, family history of SQTS, family history of sudden death at age ≤40 years, and survival of a ventricular tachycardia (VT)/VF episode in the absence of heart disease (Priori et al., 2013).

1.3.1.2.2 Brugada syndrome

Similar to LQTS, BrS, is caused by defects in the cardiac ion channels, but more specifically, the reduction in the inward movement of sodium (Na+) in the cells (Batchvarov, 2014). Patients may experience palpitations, syncope, shortness of breath, chest discomfort, nocturnal agonal breathing and VF or aborted sudden cardiac death usually occurring in the evening. The diagnostic criteria for BrS are illustrated in Figure 2. A provocation test consisting of an intravenous Na+ blocking

agent such as Ajmaline may be required if the ECG alone is not sufficient for a diagnosis (Priori et al., 2013).

Figure 2 Diagnostic criteria for Brugada syndrome (Priori et al., 2013)



Expert Consensus Recommendations on ***BrS Diagnosis***

1. BrS ***is diagnosed*** in patients with ST-segment elevation with **type I** morphology ≥2 mm in ≥1 lead among the right precordial leads V_1,V_2 positioned in the 2nd, 3rd, or 4th intercostal space occurring either spontaneously or after provocative drug test with intravenous administration of Class I antiarrhythmic drugs.
2. BrS ***is diagnosed*** in patients with type 2 or type 3 ST- segment elevation in ≥1 lead among the right precordial leads V_1,V_2 positioned in the 2nd, 3rd, or 4th intercostal space when a provocative drug test with intravenous administration of Class I antiarrhythmic drugs induces a **type I** ECG morphology.

Abbreviations: BrS-Brugada Syndrome, ECG-electrocardiogram

1.3.1.2.3 Catecholaminergic polymorphic ventricular tachycardia

The main feature of CPVT is the presence of bi-directional VT on an ECG resulting from adrenergic stimulation. This manifests clinically in the first or second decade of life, triggered by exercise or emotional stress, as a syncopal episode sometimes associated with seizure-like activity. Exercise testing on a treadmill and a 24-hour ECG recording are key diagnostic tools, apart from the resting ECG, as these will often demonstrate extra ventricular beats which then progress into biventricular or polymorphic VT during exercise (Priori et al., 2013).

1.3.1.2.4 Unexplained cardiac arrest or Idiopathic ventricular fibrillation

Idiopathic VF remains a diagnosis of exclusion following survival of an individual from cardiac arrest. The term idiopathic VF is used when systematic clinical evaluation of possible cardiac, respiratory, metabolic, and toxicological aetiologies that may lead to

cardiac arrest are normal. Ideally, VF should be documented. Long term follow up of this group revealed 21% had a specific genetic diagnosis (Visser et al., 2016).

1.3.1.2.5 Unexplained sudden cardiac death

To support the diagnosis of an unexplained SCD an autopsy as well as toxicological studies are key to exclude noncardiac causes. This should be followed by pathological evaluation with detailed histopathological examination of the heart to exclude clear causes for SCD. An ICC might be identified through these investigations such as a structural disease like HCM. However, there are cases that remain unexplained and are referred to as sudden arrhythmic death syndrome (SADS), wherein the mechanism of death could be an inherited arrhythmic syndrome (Basso et al., 2017). Family history and screening of first-degree relatives alongside molecular autopsy may be recommended in this situation (Fellmann et al., 2019).

1.3.2 Management of inherited cardiac conditions

Management in ICCs is focused on two aspects: 1. Symptom management and 2. Prevention of disease-related complications. In inherited cardiomyopathies, these will include medications such as beta blockers, calcium antagonists, angiotensin-converting enzyme inhibitors and standard heart failure therapy; and devices including pacemakers and implantable cardioverter defibrillators (ICDs) to maintain optimal cardiac function, manage heart failure and prevent sudden death. In extreme cases, cardiac transplantation may be required (Elliott et al., 2014).

In HCM, risk stratification plays an important part in prescribing device therapy for sudden death (O'Mahony et al., 2018). In obstructive HCM, LVOTO can be managed surgically if unresponsive to medical therapy, through a septal myotomy-myectomy wherein a portion of the heart muscle is removed from the intraventricular septum via

an aortic approach. This has been deemed to have better outcomes for symptom relief and need for re-intervention compared to the alternative procedure, alcohol septal ablation, consisting of an injection of 95% alcohol to produce an area of necrosis within the basal septum (Bytyci et al., 2020).

Clinical trials are currently underway for the use of a small molecule modulator of β-cardiac myosin, Mavacamten, which is thought to reduce contractility and improve ventricular compliance, in HCM patients with LVOTO. The results are promising with the Mavacamten for treatment of symptomatic obstructive hypertrophic cardiomyopathy (EXPLORER-HCM) study demonstrating improved symptoms, exercise capacity, health status, reduction of LVOTO, with benefits to cardiac structure and function (Olivotto et al., 2020).

In inherited arrhythmias, management depends on the disease sub-types. Due to the high incidence of arrhythmias in the presence of sympathetic stimulation (exercise or emotional stress) beta blockers have been the first line drugs in LQTS and CPVT. ICDs are used when medications alone are insufficient to protect against sudden death; or required on its own to prevent dangerous arrhythmias such as in BrS (Priori et al., 2013).

In all ICCs, careful discussion of tailored lifestyle advice is important because of the significant contribution to triggering signs and symptoms. Exercise advice for ICCs include modification, reduction or avoidance, particularly in HCM with LVOTO, ARVC, LQTS Type 1 and CPVT (Pelliccia et al., 2020). Patients with LQTS and BrS are given a ‘Drugs to avoid’ list as a wide range of medications, from antiarrhythmics, antipsychotics, antibiotics to anaesthetic agents, can induce QT prolongation and ventricular arrhythmias. Prompt correction of electrolyte imbalances is recommended

for inherited arrhythmias as well as rapid treatment of fever in BrS which can trigger ECG changes (Priya et al., 2015).

A specialist ICC clinic is the gold standard for providing care to ICC patients as it consists of a multi-disciplinary team of cardiologists, clinical geneticists, nurses and genetic counsellors who can give expert advice regarding the diagnosis, risk stratification, and management of the ICC, as well as providing genetic input and recommendations for the family (Burton et al., 2009, Musunuru et al., 2020).

1.3.3 Genetic aspects of inherited cardiac conditions

1.3.3.1 Genetic basis of inherited cardiac conditions

Most ICCs are autosomal dominant which conveys a 50% chance of inheritance for first degree relatives. There are rare cases of autosomal recessive, mitochondrial and X-linked ICCs (Ackerman et al., 2011). An illustrative list of the genes in ICCs are shown in Table 1.

Table 1 Illustrative list of genes in inherited cardiac conditions adapted from Musunuru et al. (2020)

Condition	Genes
Hypertrophic cardiomyopathy (HCM) **Strong evidence**	*MYBPC3, MYH7, TNNT2, TNNI3, TPM1, ACTC1, MYL2, MYL3*
Moderate evidence	*CSRP3, TNNC1, JPH2*
With syndromic features	*PLN, CACNA1C, DES, FHL1, FLNC, GLA, LAMP2, PRKAG2, PTPN11, RAF1, RIT1, TTR*
Dilated cardiomyopathy (DCM)	*TTN, LMNA, MYH7, TNNT2, BAG3, RBM20, TNNC1, TNNI3, TPM1, SCN5A, PLN* plus HCM & ARVC genes

Condition	Genes
Arrhythmogenic right ventricular cardiomyopathy (ARVC)	*DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, RYR2, SCN5A, TMEM43, TTN;* plus DCM genes
Restrictive cardiomyopathy	*TTR;* plus HCM and DCM genes
Long QT syndrome	*KCNQ1, KCNH2, SCN5A*
Short QT syndrome	*KCNH2, KCNQ1, KCNJ2*
Brugada syndrome	*SCN5A*
Catecholaminergic polymorphic ventricular tachycardia	*RYR2, CASQ2*

In inherited cardiomyopathies, pathogenic (disease-causing) variants mainly in the genes encoding sarcomeric and desmosomal proteins bring about the structural heart defects that can lead to heart failure. In DCM, nucleoskeletal, mitochondrial, cytoskeletal and calcium handling protein encoding genes have also been implicated (Girolami et al., 2018).

Inherited arrhythmias usually involve disease causing variants in the genes responsible for forming and regulating ion channels, affecting the action potential of the heart rhythm, which can lead to cardiac arrest from ventricular tachycardia or fibrillation (Chung, 2010).

1.3.3.2 Diagnostic genetic testing

The advances in genetic testing techniques have made it possible to offer diagnostic genetic testing for ICCs in the clinical setting. This involves taking a blood sample

from the proband to determine the presence of a pathogenic variant in the ICC candidate genes. Diagnostic genetic testing is typically offered to probands who fulfil the criteria for the phenotype but may also be done on a case-by-case basis for those with a high suspicion for an ICC diagnosis and collectively, genetic test results are interpreted according to standard variant classification guidelines as set by the American College of Medical Genetics and Genomics and the Association for Molecular Pathology (Richards et al., 2015). It is therefore no surprise that the yield of diagnostic genetic testing in ICCs ranges from <20% to 75% in inherited arrhythmias whilst in inherited cardiomyopathies it is <20% to 60%. Therefore, there is a high possibility that the proband will either have a negative result for a pathogenic variant or have variants of unknown clinical significance (VUS) (Musunuru et al., 2020, Tester and Ackerman, 2011).

Finding a pathogenic variant in an affected individual will usually not alter their medical management, except for certain forms of LQTS, discovery of phenocopies in HCM; and DCM accompanied by conduction disease. However, when a pathogenic variant is detected in the proband, it opens the possibility of offering predictive genetic testing (PGT) to first-degree family members who are at 50% risk of being carriers. This may also facilitate prenatal genetic testing or pre-implantation genetic diagnosis (PGD). Using these reproductive techniques will avoid passing on the pathogenic mutation to their offspring (Ackerman et al., 2011).

More recently some incidental genetic findings for ICCs are brought about when individuals who do not necessarily have a heart problem undergo diagnostic genetic testing (for example, cancer) or avail of a direct-to-consumer test which uses whole exome (WES) or genome sequencing (WGS). The American College of Medical Genetics (ACMG) has recommended a list of 59 medically actionable genes that

warrant notification for pathogenic or likely pathogenic variants even if these are not genes involved in the disease being tested for. Thirty out of the 59 genes are related to cardiovascular disease (Kalia et al., 2017). Prior to any clinical action, it is recommended that these patients are seen in a specialist ICC clinic for a comprehensive family history and detailed phenotyping. Should an ICC diagnosis arise, patients will follow a similar management pathway to that of a proband, however, many cases are asymptomatic with no phenotypic evidence for an ICC or family history (Musunuru et al., 2020). As the evidence is limited on how to progress with these cases, a tailored approach is recommended (Hershberger et al., 2018).

1.3.3.3 Predictive genetic testing

Predictive genetic testing (PGT) is when an asymptomatic individual has a genetic test to find out whether they have inherited a familial pathogenic variant. Those who test negative for the variant are typically reassured and discharged whilst those who are carriers for the pathogenic variant are kept under regular follow-up and given appropriate medical management and lifestyle recommendations (Ackerman et al., 2011)

1.3.3.4 Cardiac Screening

When the proband's genetic testing result is negative or a VUS, the presence of an ICC is not ruled out because the phenotype remains present in the proband. In this situation, at-risk family members cannot be offered PGT, and they will be invited to undergo cardiac screening to assess their risk for an ICC instead. Cardiac screening may even be offered on the outset as diagnostic genetic testing may not be readily available in some ICC centres (Ackerman et al., 2011).

Cardiac screening for a family member involves several investigations depending on the ICC of the proband. As a minimum, this will include clinical and family history taking, physical assessment, ECG, and echocardiogram. ICCs such as LQTS will require a 24-hour ECG and an exercise test on a treadmill; in BrS, an Ajmaline provocation test is added; and in ARVC cMRI is required. Relatives who have normal results are usually discharged; but still require occasional follow-up every 1-5 years, depending on their age and the proband's diagnosis. Those who show abnormalities on these tests remain on follow-up and given appropriate medical management and lifestyle recommendations. For most inherited cardiomyopathies, family members face a long period of multiple screening until they are in their mid-50s due to the possibility of late onset disease (Elliott et al., 2014).

1.3.3.5 Penetrance and variable expressivity

For a family member who is a carrier of a pathogenic variant for ICC, the risk of developing the condition is influenced by several factors, many of which are still unknown. This is the concept of reduced penetrance where it is possible that a carrier for an ICC pathogenic variant may never develop the condition. Another common feature in ICCs is that of variable expressivity where members of the same family carry the same ICC pathogenic variant and yet some are very severely affected and others only have mild signs and symptoms (Lobo, 2008).

In inherited cardiomyopathies, carriers have an increased risk of developing a structural heart abnormality but the timing and severity of this is unknown. In inherited arrhythmias, there are more practical implications of being a carrier as this increases the chances of dangerous heart rhythm problems which can cause sudden death even in the presence of normal clinical tests such as an ECG. Carriers are usually asked to adhere to a 'Drugs to avoid' list and may be prescribed a modified exercise

regime and beta-blockers (Girolami et al., 2018). It is, therefore, very likely that completely ‘healthy’ individuals are in medical care for a long-term condition which may never manifest.

1.3.3.6 Genetic counselling

Genetic counselling is the process by which patients are guided through their decision-making in genetic healthcare. The patient receives information regarding genetic risk, options to manage this risk, and supported as they adjust to this risk. ‘Non-directiveness’ is the guiding principle in providing genetic counselling and the patient has authority on deciding the next steps without coercion (Skirton, 2005). It has been argued that in conditions such as ICCs where preventative measures are available, ‘appropriate directiveness’ may be applicable (Bartels et al., 1997).

Once a pathogenic variant is found in a proband, PGT is almost always triggered for first degree family members as it is a class I indication (strong evidence or general agreement that the intervention is beneficial) in established practice guidelines, particularly for inherited arrhythmias at any age (Priori et al., 2013). Patients make multiple and sequential decisions when undergoing cardiac screening and/or genetic testing. They often feel that once the process starts, it is not easy to walk away (Emery, 2001). Therefore, a balanced approach must be maintained to preserve autonomy. It is recommended that the genetic counselling session should include an exploration of the patients’ coping mechanisms, family dynamics, psychological and emotional state to determine the possible impact of a carrier or non-carrier result and how might clinicians provide support (Ingles et al., 2011).

1.3.3.7 Impact of inherited cardiac conditions

A diagnosis or carrier status for an ICC will bring with it profound consequences for an individual and their family. Not only are they subjected to the complex diagnosis and management pathways undertaken in the context of preventing sudden cardiac death, but the genetic nature of ICCs also requires the task of simultaneously dealing with their own health whilst addressing the risk for their family. This situation brings forth a whole host of psychosocial issues known to patients affected by genetic conditions (McAllister et al., 2007).

Focusing on the care of at-risk relatives for ICCs, the condition is different to other genetic conditions like Huntington disease due to the availability of preventative measures. The cost of a single preventable acute event in HCM in 2007 was estimated at £20,000 (Wordsworth et al., 2010) and prompt management with an ICD if appropriate has been proven to be cost effective both in terms of saved lives and quality adjusted life years (Magnusson and Wimo, 2020). Hence, there is an opportunity to decrease costs through prevention. This does not negate the need for family members to be aware of the implications of a negative or positive result. Even if they are asymptomatic or have very few symptoms, they may still be subjected to radical lifestyle changes or interventions to reduce the risk of sudden cardiac death. This includes exclusion from competitive sport, which for some could be an important feature in their life (Asif et al., 2015, Pelliccia et al., 2020) or recommendation for an ICD implant, which has been linked to anxiety, depression across all groups of cardiovascular patients (Jackson and Murphy, 2017, Shiga et al., 2013).

Facilitating effective communication of risk information relating to the ICC is essential and the way a family functions can influence this. In ICCs, it is reported that 20-40% of relatives are either unaware of their genetic risk or do not act on this knowledge

(Burns et al., 2016, Christiaans et al., 2008, Gaff et al., 2007). It is well documented that the family unit can change profoundly in the context of a serious, chronic medical illness (Gonzales et al., 1989) and communication breakdown is not uncommon within families affected by genetic disorders (Rowland and Metcalfe, 2013, Ormondroyd et al., 2014). It is, therefore, no surprise that there remains poor uptake of screening and/or PGT in ICCs despite availability of effective management (Department of Health, 2013, Christiaans et al., 2008, van den Heuvel et al., 2020).

Guidelines exist on the care of family members at risk for ICCs (Elliott et al., 2014, Musunuru et al., 2020, Priori et al., 2013) which all recommend an attendance at a specialist multi-disciplinary specialist cardiovascular genetics clinic. The clinics aim to provide comprehensive care, focusing not just on the medical and genetic aspects of ICCs but also providing psychological support (Watts et al., 2009, Caleshu et al., 2016). The demand is already exceeding the capacity of clinics in existence in the UK (Burton et al., 2010). However, specialist services are growing in number with a better geographic spread (Stephenson, 2017). In our own centre's experience, within a period of 10 years, we have grown from two clinics per week with 10 patients per clinic to seven clinics per week with 12-15 patients per clinic and provide a regional service (Bueser, 2017). With increased public awareness, improved screening and genetic testing techniques and the push for mainstreaming genomics as government policy (HMGovernment, 2020) this will bring about the need for more effective and evidence-based means of providing support as patient numbers increase in an area where health services are already stretched.

1.4 Psychoeducational interventions

Providing psychoeducational interventions is an established way of providing a therapy to facilitate coping with an experience related to an illness through instructive

material and therapeutic communication techniques (Chan, 2005). Psychoeducation may have multiple components that combine patient education with the provision of counselling, behavioural change techniques and social support. They can be delivered in a variety of settings including hospitals and community settings, via correspondence, telephone or online (Barsevick et al., 2002, Donker et al., 2009).

1.4.1 Psychoeducational interventions in cardiovascular and genetic healthcare

In patients with cardiovascular disease, meta-analyses of psychoeducational interventions reveal that they are effective in reducing chest pain (McGillion et al., 2014) alongside improving quality of life (QoL) (McGillion et al., 2008); and improve physical activity levels (Aldcroft et al., 2011, McGillion et al., 2014), facilitate smoking cessation (Huttunen-Lenz et al., 2010) and reduce overall mortality (Dusseldorp et al., 1999). Furthermore, a Cochrane systematic review found that psychological interventions for patients with coronary heart disease improved psychological symptoms and reduced cardiac mortality with the caveat that there is uncertainty of the extent of these improvements and the need to tailor techniques to specific issues experienced by patients (Richards et al., 2018).

In genetic healthcare, psychoeducational interventions have been implemented mostly in hereditary breast cancer, mainly within the setting of genetic counselling or living with the increased risk of the condition. These have been found to be acceptable (Halbert et al., 2004, Katapodi et al., 2018), useful in improving knowledge and allaying stress (McKinnon et al., 2007, Roussi et al., 2009, Maheu et al., 2015, Appleton et al., 2004) and helpful in increasing uptake of genetic counselling (Kasting et al., 2019).

A psychoeducational intervention to be used across genetic conditions has also been developed based on the multi-family discussion group (MFDG) model to support the family as a unit to facilitate communication, assist in adaptation and coping with the condition. So far, it has been proven to be acceptable to patients and feasible for delivery by genetic counsellors and is awaiting a definitive clinical trial (Eisler et al., 2016, Eisler et al., 2017).

1.4.2 Psychoeducational interventions in ICCs

In the care of patients with ICCs, psychoeducational interventions are only just being developed. One study (Hodgson et al., 2016) has been published on a genetic counselling intervention for ICC probands at the time of diagnosis. This was developed based on the Reciprocal Engagement Model of Genetic Counselling (REMGC) which aims to incorporate both educative and counselling aspects in a client-counsellor interaction. The aim was to help facilitate communication of risk to family members. A randomised control trial (RCT) comparing the intervention which consisted of three additional telephone calls by a genetic counsellor versus routine practice (no additional contact) showed that there was no significant difference in at-risk relatives contacting genetic services. However, it was noted that sub-group analysis revealed a significant increase in high-risk relatives (first degree relatives as opposed to more distant kinship) seeking a referral with the intervention. A limitation of this study was how outcome data was collected which resulted in the underestimation of the uptake of contact with genetic services which, at most, was only 25.6% of eligible relatives.

An RCT for a genetic counsellor-led custom-designed communication aid for relaying ICC genetic results to probands has been conducted (Smagarinsky et al., 2017). The intervention aimed to increase the confidence of the proband in relaying genetic test

results to their at-risk relatives; and improve their genetic knowledge and adaptation to genetic information. This consisted of a face-to-face consultation with telephone follow-ups at one, three and six months. (Burns et al., 2019). Twenty-two probands were randomised to the intervention arm and 20 to the control arm. There were no statistically significant differences in the outcomes, however, there was a trend for genetic knowledge scores consistently higher amongst the intervention group. Worryingly, despite the intervention and the setting of this study in a specialist ICC clinic, up to 29% of at-risk relatives remain uninformed about a genetic result in their family and up to 17% of at-risk relatives remain uninformed of the HCM diagnosis itself (Burns, 2019).

A major limitation in Burns' (2019) RCT is that it was underpowered. An RCT has been proposed with similar aims but this time with a bigger sample size of probands with genetic results (total n=85) and within a multi-centre setting. The participants will be randomised to the intervention, which will be a tailored approach wherein the genetic counsellor and the proband decide together to either inform relatives of their risk directly through a genetic counsellor or through the proband, or allocated to usual care wherein relatives are informed by the proband only (van den Heuvel et al., 2019). No results have been reported at the time of writing.

Another study in progress is the DCM Precision Medicine Study which, apart from providing exome sequencing to 1300 patients with DCM and cardiac screening to 2600 at-risk relatives, will assess the effectiveness an intervention called Family Heart Talk, to aid family communication, for improving uptake of preventive screening and surveillance in at-risk first-degree relatives (Kinnamon et al., 2017). The Family Heart Talk intervention is a guide to family communication about DCM available in both print and web-based forms. Visuals and lay language explanations of the care of

individuals with DCM, including the necessity of cardiac screening in family members are included. Guidance on talking with family members about DCM risk with samples of e-mails and letters are also available.

Two RCTs have been published on a psychoeducational intervention which mainly involves mindfulness-based stress reduction (MBSR) training for a mixed group of patients with a cardiac diagnosis, which included patients with cardiomyopathy and LQTS (Gotink et al., 2017, Freedenberg et al., 2017). In Gotink et al. (2017), there was no indication of what type of cardiomyopathy and if this was inherited in the 84 intervention participants and 31 control participants (usual care) diagnosed with cardiomyopathy out of the 324 participants. The overall results for all participants were that the 12-week online MBSR intervention showed small but significant trends for improving exercise capacity, systolic blood pressure, mental functioning and depressive symptomatology after a one-year follow up period.

Freedenberg et al. (2017) compared a six-week group-based MBSR intervention with a video online support group in a mixed group (N=46) of adolescents and young adults with a cardiac diagnosis. There was a total of ten patients with unspecified cardiomyopathy and LQTS in both groups and it was not discernible if they were part of the 18 patients who had pacemakers or ICDs. Both the MBSR intervention and video support groups reported reduced stress. Whilst these two RCTs sound promising in supporting ICC patients with the psychosocial impact of their condition, both studies included poorly described cohorts of ICC patients. Furthermore, measures of efficacy were not consistently based on clinical cut-offs, only improvements from baseline measures which makes it is difficult to ascertain clinically meaningful changes.

Psychoeducational interventions are yet to be fully explored in ICCs. By building upon those described in cardiovascular and genetic healthcare and the six studies involving ICCs, there is a potential to develop psychoeducational interventions that are feasible, acceptable, and beneficial patients.

1.5 Justification for the thesis

Current psychoeducational intervention and clinical service development in ICCs focus primarily on engaging the proband to communicate with first degree relatives to facilitate cardiac screening and/or PGT. This is an important strategy in family care for the prevention of significant morbidity and sudden cardiac death especially as a recent long-term follow-up study showed that only 60% of at-risk family members seek genetic counselling (van den Heuvel et al., 2020). Furthermore, it has also been reported that 25% of known carriers for ICCs drop out of follow up (Christiaans et al., 2009b). Therefore, if because of their experience, these family members, the majority of who are asymptomatic, end up with disruption of family dynamics, psychosocial stress leading to physical decline or non-adherence to medical advice, then the preventative aim is defeated. There will also be grave consequences for the health of subsequent family members if this patient group is not supported to facilitate ongoing genetic risk communication.

To date, there are no psychoeducational interventions focused on supporting at-risk family members as they undergo the process of cardiac screening and/or PGT for ICCs. The short and long-term needs of this group have yet to be comprehensively reviewed to facilitate the development of appropriate supportive interventions as they go through this process and deal with the outcomes. Given the speed of technological advances which enable faster diagnosis in ICCs and increased accessibility of genetic testing within the setting of extremely busy clinical services, it is essential that

innovative evidence-based psychoeducational interventions are developed for this growing population to support optimal health, informed decision-making, timely adjustment to health status and maximal coping strategies.

1.6 Structure of the thesis

This thesis is a report on the development of a psychoeducational intervention focused on the care and support of at-risk family members for ICCs. This section describes the organisation of the thesis following on from the Introduction chapter.

1.6.1 Research design

Chapter 2 sets out the overall aims and objectives of the thesis and describes the overarching research design of the project based on the Medical Research Council (MRC) framework for developing and evaluating complex interventions (Craig et al., 2008). This project is focused on the Development stage of the MRC framework: identifying the evidence base, identifying appropriate theory, and modelling process and outcomes. The chapter also provides an overall description of the series of studies undertaken to meet the corresponding aims and outputs as stipulated within each applicable aspect of the MRC framework and how these were tailored for the project. The individual studies are reported in full detail in later chapters.

Patient engagement was central to the planning and conduct of this project, therefore the roles and responsibilities of the KHP-PPIICC group is discussed in this chapter alongside the ethical considerations in the research process.

1.6.2 Phase 1: Identifying the evidence base

In establishing the evidence base of the psychoeducational intervention, a systematic review was conducted. Chapter 3 details a mixed methods systematic review

conducted to obtain a comprehensive view of the experiences of at-risk family members as they undergo cardiac screening and/or PGT for ICCs.

1.6.3 Identifying and developing theory

Chapter 4 presents a critical analysis of the theoretical frameworks that guide the practice of delivering genetic healthcare, with a focus on genetic counselling theories and decision-making. There is also a thorough discussion and justification of the appropriate theory selected to underpin the psychoeducational intervention.

1.6.4 Phase 2: Developing the intervention model

A qualitative study consisting of group and individual interviews is reported in Chapters 5-7 with a view to determine the psychoeducational intervention model. This study explored the experiences and psychosocial needs of patients who had undergone cardiac screening and/or PGT and sought their views on a psychoeducational intervention designed to address these needs and those identified from the systematic review.

1.6.5 Phase 3: Modelling process and outcomes

Chapter 8 describes how the overall design of the psychoeducational intervention was developed by synthesising the evidence gathered in the preceding studies and the consensus process undertaken by the KHP-PPICC group. This chapter also includes a critical analysis of the design of the intervention model and mapping of components, delivery, and outcome measures to the selected theoretical framework.

1.6.6 Discussion and conclusions

Chapter 9 brings together the results from the series of studies described and provides a discussion in the context of similar studies and the fast-moving field of

cardiovascular genetic healthcare. This chapter also contains a critique of the methodology and limitations of the project, as well as implications of the findings for clinical practice and further research. The final conclusive statements are made, and a list of dissemination activities are listed, completing the thesis.

Chapter 2: Research design

2.1 Introduction

This chapter provides an overview of the research design applied to the PISICC project which consisted of a series of research studies based on the MRC framework for designing complex interventions (Craig et al., 2008). The aim and objectives of the PISICC project are stated in section 2.2. In section 2.3, the methodology is described incorporating the principles of complex intervention development followed by a discussion of the MRC framework and the role of PPI. The use of mixed methods in the inter-related studies that comprise the PISICC project are discussed within the context of how they address the three aspects of the Development stage of the MRC framework (Identifying the evidence base, identifying/developing theory; and modelling process and outcomes). Methods for the individual studies are reported in greater detail in later chapters (Chapters 3-5). In the final section, the ethical considerations throughout the conduct of this study are discussed.

2.2 Aims and Objectives

2.2.1 Overall aims

The purpose of the PISICC project is to develop an evidence-based psychoeducational intervention to address the needs of patients who have a new diagnosis or carrier status for an ICC following cardiac screening and/or PGT. The aims of the overall project were to:

- Develop a healthcare intervention incorporating patient education and psychological support.
- Optimise the intervention model according to the preferences of end-users.

2.2.2 Objectives

The specific objectives to achieve the overall aims were as follows:

- To establish the evidence base for the psychoeducational intervention for patients with a new diagnosis or carrier status for an ICC following cardiac screening and/or predictive genetic testing.
- To establish the theoretical basis of a psychoeducational intervention within the context of genetic healthcare.
- To determine the components and features essential to the intervention model.
- To identify primary and secondary outcomes associated with the intervention.
- To incorporate the perspectives and preferences of the end-users in the intervention model.

2.3 Methodology

2.3.1 Research design

Interventions are deemed to be any action by a health professional with an aim to improve the situation of an individual with health or social care needs (Richards and Hallberg, 2015). The intervention is termed ‘complex’ when there are multiple interacting components (Craig et al., 2008). It has also been recognised that complex interventions also often involve many factors that need to be accounted for such as outcome measures, organisational context and evaluation (Datta and Petticrew, 2013). The psychoeducational intervention being developed within the PISICC project

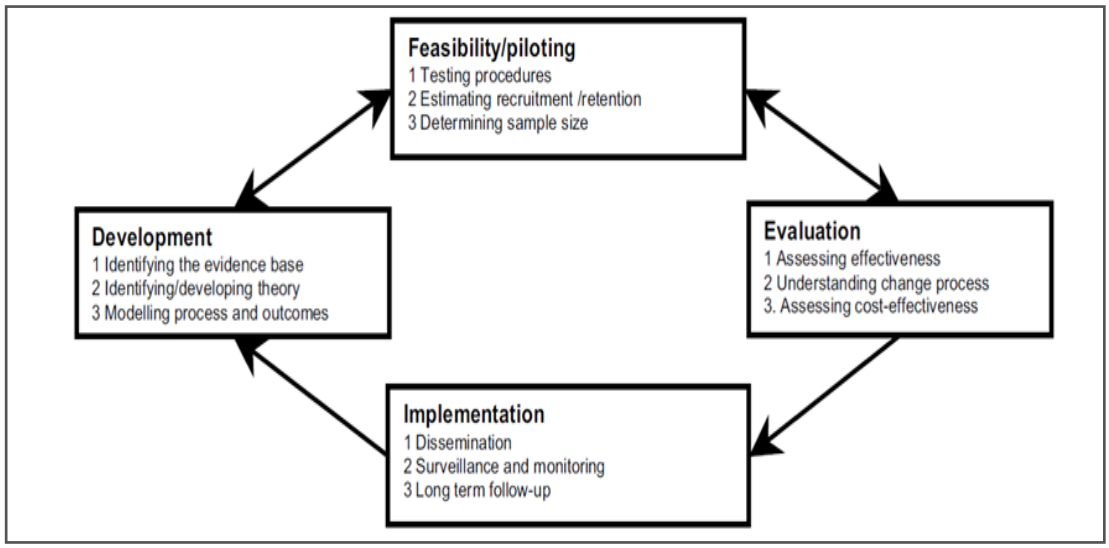
is considered complex due to the multiple components for patient education, support for coping and management strategies, and peer support.

Despite an emphasis on rigour when developing interventions, it has been noted that many never impact on clinical practice and become ‘research waste’ (Chalmers et al., 2014). To avoid this phenomena in complex intervention development, guidelines have been proposed to provide a systematic approach from the point of recognition of a healthcare need to the establishment of a therapeutic solution (Conn et al., 2001, Craig et al., 2008, van Meijel et al., 2004, Whittemore and Grey, 2002) . The common principles within these guidelines specify that a complex intervention must be: evidence-based, have a strong conceptual/theoretical basis; and are acceptable and feasible for use in the target population within the context of routine practice (Bleijenberg et al., 2018b).

The MRC Framework for the development and evaluation of complex interventions was chosen as the overall research design for the PISICC project as it is an established process that not only adheres to the principles mentioned above but also follows an iterative development approach. The MRC framework specifies a development-evaluation-implementation process which consists of four stages that is summarised in Figure 3 (Craig et al., 2008). As opposed to a linear-stepwise approach, the MRC Framework implies that there are several interactions between the initial Development stage and the Feasibility/piloting stage; as well as the Evaluation stage before an intervention is brought forward to the Implementation stage. These built-in feedback loops emphasise the reciprocal relationships between components of the intervention, continuously allowing for adjustments as evidence

arises so that only a robust intervention model goes through to a definitive clinical trial (Bleijenberg et al., 2018b).

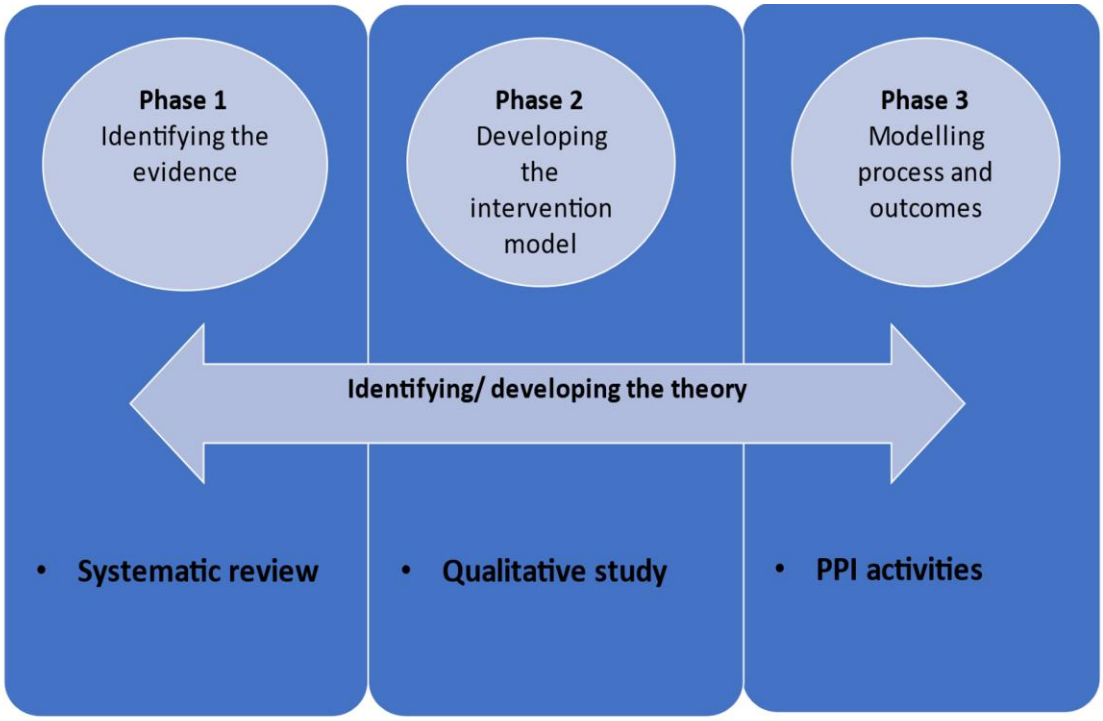
Figure 3 Stages of the Medical Research Council Framework for the development of complex interventions (Craig et al., 2008)



The initial stage in the MRC Framework is the Development stage which consists of three steps: Identifying the evidence base, Identifying/developing theory and Modelling process and outcomes. Thorough investment in this stage avoids 'research waste' and minimises the risk of participants being subjected to ineffective or unacceptable interventions (Chalmers et al., 2014). The scope of the PISICC project is focused on this aspect of intervention development as it is arguably the most crucial stage and has implications for the success of both the evaluation and implementation stages.

In order to address the aims and objectives of the PISICC project, a series of individual but related studies were devised, informed by a patient and expert advisory group (INVOLVE, 2012). This mixed methods multi-phase approach allowed for each sequential quantitative and qualitative study to build upon the learning from previous knowledge (Cresswell and Plano Clark, 2017). The scope of the thesis and an overview of the studies conducted within the Development stage of MRC Framework are summarised in Figure 4.

Figure 4 Overview of the studies conducted within the Development stage of Medical Research Council Framework for the development of complex interventions



Abbreviation: PPI-Patient & public involvement

2.3.2 Patient and public involvement

Lack of PPI in research has also been cited as a cause of 'research waste' and this must go further than contributing to the research question (Chalmers et al., 2014) . Acceptability, feasibility and appropriateness to patients and clinicians are key to the adoption of complex interventions in clinical practice (van Meijel et al., 2004). At the

inception of the PISICC project in 2015, a formal consultation and review of the proposal was conducted by five members of the Cardiovascular PPI group organised by the Biomedical Research Centre User Involvement Team of King's Health Partners (KHP). Further opinions were sought from five members of the patient support groups Sudden Arrhythmic Syndrome UK (SADS UK) and Cardiomyopathy UK. This consultation resulted in strengthening the case for the conduct of the research as it was considered relevant and appropriate for the patient group, as well as the use of mixed methods to draw on patient experiences to ensure the applicability of findings.

At the commencement of the PISICC project in June 2016, the King's Health Partners' Patient & Public Involvement for Inherited Cardiac Conditions (KHP-PPIICC) group, was formally established according to INVOLVE guidelines (INVOLVE, 2012, INVOLVE, 2013) and have provided input all throughout the planning and conduct of the research. The terms of reference for the group can be found in Appendix 1. The KHP-PPIICC group was comprised of patients and family members affected by ICCs and expert ICC clinicians to ensure that the intervention is appropriate and of maximum benefit to patients. The group consisted of ten adult patients and family members recruited from the ICC clinic and the Cardiomyopathy UK support group. It is a diverse group with not one ICC entity dominating. In addition, three clinicians joined the group including a cardiologist, geneticist, and cardiac genetic nurse. Aside from face-to-face meetings, contact via e-mail was maintained during the duration of the research and the group will continue to participate in the dissemination of research results.

The KHP-PPIICC group also played a major role in the modelling of the intervention and outcomes which will be described further in Section 2.3.3.4 and Chapter eight.

The role of the group is faithful to the pragmatic philosophical approach of the research and their contributions are invaluable to the outputs herewith.

2.3.3 The scope of the PISICC project within the MRC framework

The MRC Framework for complex interventions was utilised to fulfil the essential elements for developing the psychoeducational intervention and to address the overall aims and objectives. The PISICC project encompassed the three aspects of the Development stage of the MRC framework incorporating a mixed methods approach in the series of studies conducted within each phase.

2.3.3.1 Phase 1: Identifying the evidence base

To ensure a solid basis for developing a psychoeducational intervention to support patients who have a new diagnosis and/or carrier status for an ICC following cardiac screening and/or PGT, evidence on existing interventions for this population was sought. However, as mentioned in Chapter 1, there are only a few psychoeducational interventions developed in ICCs and they are mainly focused on probands rather than the at-risk family members. Whilst it is important to note the findings of these studies, due to the lack of existing established interventions for this population, the starting point was to have a greater understanding of the experiences of at-risk family members as they undergo the cardiac screening and/or PGT process (Richards and Hallberg, 2015).

A mixed methods systematic review was undertaken according to the Centre for Reviews and Dissemination methods for undertaking reviews in health care

(University of York NHS Centre for Reviews Dissemination, 2009) to gain a comprehensive view on the experiences and preferences of adult patients at risk for ICCs as they undergo cardiac screening and/or PGT, including the psychosocial impact of this process, and to identify areas requiring further support and development in the care pathway (full details are reported in Chapter 3).

2.3.3.2 Identifying/developing the theory

In developing interventions, identifying and developing an underpinning theory is of vital importance to elucidate the mechanism of action and impact of the proposed intervention (Datta and Petticrew, 2013). Chapter 4 provides a discussion of existing and emerging theories in genetic healthcare and the justification for the theory selected.

The findings of the systematic review brought about themes surrounding the experiences, psychosocial impact, challenges, and aspirations of patients undergoing cardiac screening and/or PGT and thus, identified important areas for the intervention to target. Incorporating theory in developing the PISICC intervention has facilitated the understanding of the mechanism of how and why these themes emerged bringing forth a theoretical framework from which to develop the intervention (Bleijenberg et al., 2018b)

Building on the evidence from Phase 1 and the application of theory, a qualitative study was conducted to identify the components and preferences for the PISICC intervention. The theoretical framework provided an important feedback loop to check

for consistency and gaps in evidence between the systematic review and the qualitative study (Figure 4); and was therefore essential to evidence synthesis (Davis et al., 2015). Furthermore, in order to elicit the practical aspects of designing the intervention, the theoretical framework guided the identification of variables which should be targeted for manipulation by the intervention and the accompanying outcome measures (Eccles et al., 2005). The identification and development of the theoretical underpinnings of the intervention impacts the breadth of the Development stage of the MRC Framework as depicted in Figure 4 and will be referred to throughout the phases of the study.

2.3.3.3 Phase 2: Developing the intervention model

Building on the evidence from the systematic review from Phase 1 of the PISICC project, the qualitative interviews with family members who have undergone cardiac screening and/or PGT were conducted to explore their experiences of the screening/PGT process; and views on a psychoeducational intervention-the components, design and delivery considerations (Richards and Hallberg, 2015). This study also provided an opportunity to test the applicability of the theoretical framework identified (Bleijenberg et al., 2018b) and corresponded to the feedback loop in the incremental approach of the MRC Framework. This phase was essential in eliciting an intervention model rooted in a contemporary context with the target end-users (Craig et al., 2008). A full report on the qualitative study can be found in Chapters 5-7.

2.3.3.4 Phase 3 Modelling process and outcomes

In the final step of the Development stage of the MRC Framework, the active components of the intervention are modelled by synthesising the evidence gathered from Phase 1 and 2 of the PISICC study (Craig et al., 2008). This is a cumulative process wherein previous knowledge provides insight into the modifiable variables that influence the delivery of the intervention, the extent to which variables can be manipulated and how strongly related the variables are with the outcomes (Bleijenberg et al., 2018b). Following the principles of integration in mixed methods which recommends a joint display of the findings, the synthesis of the systematic review and the qualitative study are presented in a logic model (Cresswell and Plano Clark, 2017, Richards and Hallberg, 2015).

A logic model is a diagram which maps out an intervention and the proposed links between the intervention and the expected outcomes to summarise a theory of how an intervention might work (Anderson et al., 2011). Logic models have been recommended as a useful method of synthesising and describing the complexity within interventions (Baxter et al., 2014). Presenting a logic model allows for an easy visualisation of a) the underlying theory and assumptions of causality between the intervention and intended outcomes b) specifying the intervention components and their inter-relationships c) depicting the context within which the intervention will be implemented and their interactions (Rohwer et al., 2017).

The logic model allowed the presentation of the prototype of the PISICC intervention model to the KHP-PPIICC group where a modified nominal group technique was used to obtain a consensus on the prototype and the outcome measures identified (Jones

and Hunter, 1995). The full details of the modelling phase are in Chapter 8 which includes the logic model and prototype for the PISICC intervention.

2.4 Ethical approval

The qualitative study in Phase 2 of this project required specific ethical approval which was granted by the London-Fulham Research Ethics Committee (REC) on January 16, 2017 (reference: 17/LO/0059). Permission to conduct the study was granted by the NHS Health Research Authority on January 31, 2017.

2.4.1 Ethical considerations

The purpose of conducting clinical research is to form conclusions and/or generalisations from a systematic method of data collection and analysis to improve clinical practice and for patient benefit (Orb et al., 2001). As such, the conduct of the PISICC project adhered to core ethical principles to ensure the integrity of the scientific methods used and protect the welfare of all participants (Heale and Shorten, 2017). Issues surrounding the ethical principles of informed consent, minimising harm and confidentiality which are pertinent to this study are discussed.

2.4.1.1 Informed consent

Prospective participants should have comprehensive information about the research to enable them to make an informed and autonomous decision on their possible involvement. The study poster, patient information leaflet and consent form in Appendix 2 for the PISICC study were written in plain English and were evaluated by the REC and the KHP-PPIICC group. Within these documents, full details of the

reasons why they have been approached, the purpose of the study, the activities to be undertaken including the discussion of sensitive topics, how and where the information they provide will be used and stored; and where to find more information and contact details for questions and issues. It was also explicitly stated that they could withdraw their consent to participate in the study without giving a reason. Furthermore, potential participants were given at least 48 hours to consider joining the study and were encouraged to ask for clarifications over the telephone or face-to-face.

2.4.1.2 Protecting anonymity and confidentiality

Throughout the conduct of this study, utmost care was taken to preserve participant anonymity and confidentiality and to comply with existing guidelines including the Caldicott Principles (Department of Health, 2003) and the General Data Protection Regulation (Information Commissioner's Office, 2018). The measures taken include removal of all identifying data in the qualitative study transcripts and the use of unique study IDs and pseudonyms for participants, and secure transfer and storage of written and electronic information.

For the qualitative study, the patients who preferred one-on-one interviews were interviewed in their homes or in a private room within King’s College London (KCL). For those who participated in group interviews, some family groups were interviewed in their homes and mixed groups were interviewed in a private room at KCL. I anticipated that it would be impossible to preserve anonymity and confidentiality amongst the participants in groups, however, ground rules were explained before the

session commenced regarding the participants' responsibility for confidentiality of the information shared within the group.

2.4.1.3 Minimising harm

The welfare of the participant is first and foremost in the conduct of research and strategies for mitigating risk and harm were considered and put in place (Heale and Shorten, 2017). In the qualitative study, participants who did not want to discuss sensitive issues in a group or if it was more convenient, were given the option to have an individual interview. In the group discussions, participants had varied backgrounds, experiences and outcomes and they may not have previously encountered the discussion points in the topic guide therefore, there was a risk of coming across issues or information that they may have not considered in terms of their genetic risk. During the research, two participants needed clarification on certain aspects of their care and were directed to their clinical team to address these concerns. One group interview participant became tearful when she was sharing her experiences and was reassured that she did not have to continue, however, after a short pause, she decided that she would like to carry on. The revelation of substandard care during the study did not occur, however, a provision for addressing this was in place.

The time to participate in this study may pose a burden to the participant, therefore reimbursement for travel and childcare was provided as well as refreshments during the focus group session or interview (INVOLVE 2013).

Chapter 3: Phase 1 Identifying the evidence base

3.1 Introduction

This chapter reports on Phase 1 of the PISICC project which is the systematic review of the experiences, impact, and preferences of family members at risk for an ICC undergoing cardiac screening and/or PGT. This corresponds to the first step in the MRC framework in developing complex interventions (identifying the evidence base). The background and justification for the review is given in Section 3.2. The systematic review method which allows for the comprehensive evaluation and synthesis of different types of research is discussed in Section 3.3. The results are presented in Section 3.4 and the ensuing discussion in 3.5. The overall conclusions and recommendations for the intervention are presented in Section 3.6 and 3.7, respectively. A discussion of current evidence is also presented in a later chapter within this thesis (Chapter 9).

3.2 Background and justification

A typical starting point in the MRC Framework to identify the evidence base for a complex intervention is to look at the effectiveness of existing interventions for the target population (Craig et al., 2008). This can establish whether a new intervention is needed, and if so, identify where the gaps are in service provision, and any barriers or facilitators in implementation. A systematic review of the intervention may already exist or researchers may undertake one to establish a solid evidence base (Richards and Hallberg, 2015). As stated in Chapter 1, psychoeducational interventions have been developed for patients and families affected by cardiovascular or genetic conditions; or when these 2 conditions are combined in an ICC. However, at the time of writing, there are no psychoeducational interventions reported in the published

literature specifically developed for the first-degree relatives of ICC probands undergoing cardiac screening and/or PGT.

Although psychoeducational interventions have not been developed for at-risk first-degree relatives, there is a growing narrative on their characteristics and experiences that justify the need to develop supportive mechanisms specifically for them (Aatre and Day, 2011, Caleshu et al., 2016). Indeed, provision of genetic counselling and attendance at a specialised multi-disciplinary clinic are recommended by clinical guidelines as measures to ameliorate the impact of an ICC diagnosis or carrier status (Charron et al., 2010, Priori et al., 2013), however, how these are delivered and experienced by patients have not been systematically studied. Therefore, there was a need to establish the baseline evidence rooted in the patient experience from which to draw on to develop the psychoeducational intervention (Richards and Hallberg, 2015). This facilitated identification of areas requiring further support and development in the care pathway for which interventions can be targeted.

3.3 Review purpose, aim and objectives

The purpose of conducting this systematic review was to establish the evidence base on which a psychoeducational intervention can be developed, to support at-risk relatives as they undergo cardiac screening and/or PGT for ICCs.

The aim of this systematic review was to explore the experiences and preferences of adult patients at-risk for ICCs when they undergo cardiac screening and/or PGT. More specifically, the review objectives were the following:

- Describe how family members at-risk for an ICC experience the care provided in the healthcare setting in the context of cardiac screening and/or PGT.
- Determine the psychosocial impact of cardiac screening and/or PGT.

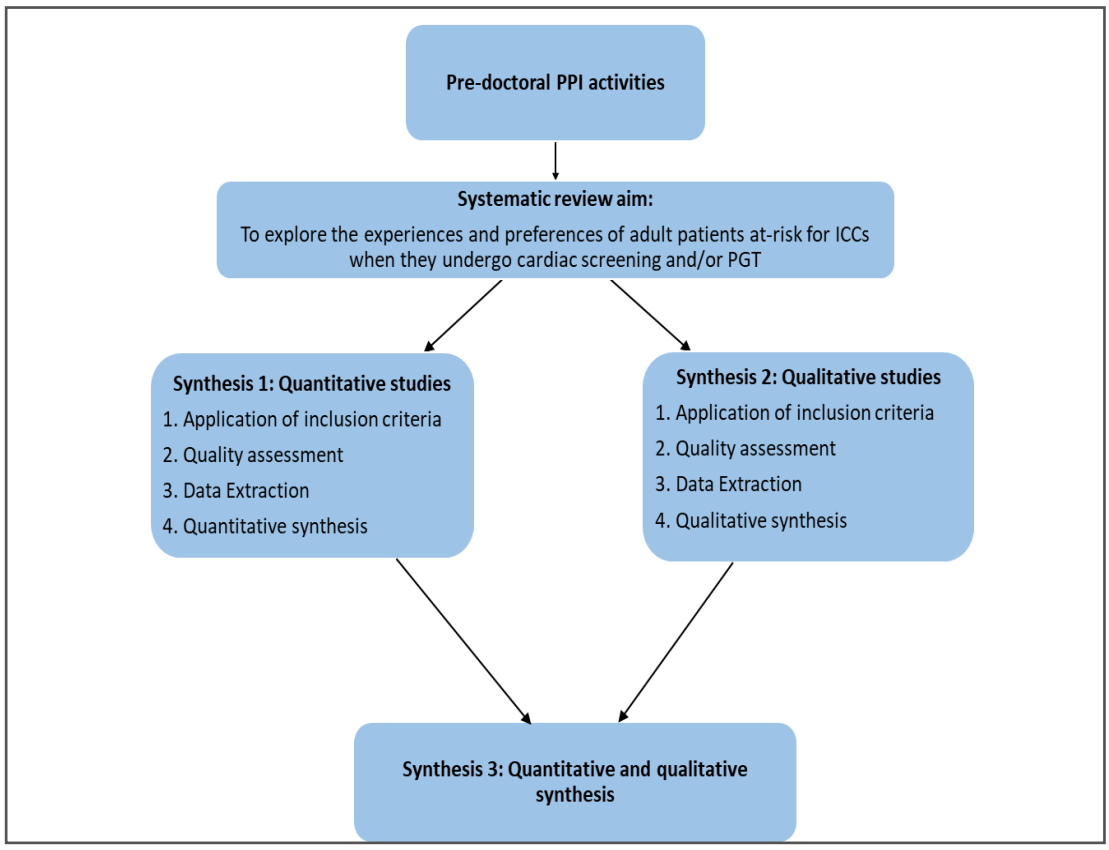
- Based on patient experience, identify the preferred options for care delivery.
- Identify the unmet support needs of this patient group.

3.4 Method

A systematic review utilises an explicit method to search, critically appraise and synthesise a body of evidence on a specific topic (Akobeng, 2005). Traditionally, systematic reviews have favoured quantitative evidence in the form of results from randomised control trials to answer questions regarding clinical effectiveness (Higgins and Green, 2009). However, in areas such as exploring patient experiences and impact of an intervention, including qualitative evidence can provide a greater understanding of contextual factors which brings forth outcomes that are valued by patients and their families (Dixon-Woods et al., 2004). Mixed methods reviews have, therefore, become increasingly utilised in healthcare to assist in a greater understanding of quantitative evidence or to provide corroboration of findings obtained from quantitative and qualitative data (Hong et al., 2017).

In order to achieve the aims and objectives of this review, the guidance according to the Centre for Reviews and Dissemination (CRD) methods for undertaking reviews in health care (University of York NHS Centre for Reviews Dissemination, 2009) was adopted to allow for a wide coverage of literature pertinent to the experiences of at-risk family members undergoing cardiac screening and/or PGT for ICCs. Thematic synthesis (Harden and Thomas, 2005, Thomas and Harden, 2008) of the results was carried out by conducting a parallel synthesis of the quantitative and qualitative data, then followed by a third synthesis combining both (Figure 5).

Figure 5 Systematic review data synthesis



Abbreviations: ICC-Inherited Cardiac Conditions, PGT-Predictive Genetic Testing, PPI-Patient & Public Involvement

3.4.1Search strategy

The search was focused on identifying published studies written in English that reported the experiences and preferences of family members at risk for ICCs as they underwent cardiac screening and/or PGT; and the impact of this on their lives. The following electronic databases indexing medical and psychosocial research were searched for papers as listed below:

- Allied and Complementary Medicine Database (AMED), 1985 to 2017 Week 26 (via Ovid)
- Embase, 1974 to 2017 Week 26 (via Ovid)
- Medline, 1946 to 2017 June 28 (via Ovid)

- PsychINFO, 1806 to 2017 June week 3 (via Ovid)
- Cumulative Index of Nursing Allied Health Literature (CINAHL) 1919 to 2017 June 28
- Pubmed earliest to 2017 June 28
- Cochrane CENTRAL earliest to 2017 June 28

The electronic databases were searched from the earliest time available to June 2017. The reference list of all identified reports and articles were also searched for additional studies. Variations and Boolean connectors of the keywords were used in the literature search. The keywords included were: family; predictive; presymptomatic; genetic; counselling; cascade; testing; screening; inherited; heart; cardiac; cardiovascular; conditions; arrhythmia; long qt; brugada; catecholaminergic polymorphic ventricular tachycardia; syndrome; cardiomyopathy; hypertrophic; dilated; arrhythmogenic right ventricular.

Appendix 3 is an example of a search strategy used in Medline (Ovid).

3.4.2 Inclusion and exclusion criteria

The inclusion criteria for this systematic review included studies that were:

- Published in English.
- Empirical peer-reviewed research articles using qualitative, quantitative, mixed, and participatory methods.
- Those that included adult (16 years old and above) at-risk patients or family members undergoing predictive genetic testing and/or cardiac screening for ICCs.
- Focused on the perceptions, experiences, preferences, actions, and strategies of this patient group.

Papers were excluded from the review if:

- There was an inability to obtain a full text version of the article.
- Focused solely on the perspectives of patients already diagnosed with an ICC and who are on a risk stratification and management pathway.
- Targeted only towards the perspectives of healthcare professionals; and yield of screening and/or PGT.
- They were guidelines for testing.

3.4.3 Selection of studies and quality appraisal

Citations retrieved from the initial search were transferred to EndNote7™ and duplicates were removed. The titles and abstracts of papers identified were screened independently by two review authors (CP and TB) against the inclusion criteria. At this stage, papers were rejected if titles and abstracts indicated that they were not in English, not original research and did not focus on the topic and population.

Studies that met the inclusion criteria underwent independent quality appraisal by two authors using the 'QualSyst' quality assessment criteria (Kmet and Lee, 2004) which can be used for both qualitative and quantitative evidence across a wide range of study designs. Points are given to indicate the extent to which specific aspects of the methodology and reporting of results are met and marked as 0 points (not addressed), 1 point (partially addressed) or 2 points (satisfactorily addressed). The summary score was calculated by dividing the total score of all applicable items by the highest possible score after excluding non-applicable items (Kmet and Lee, 2004). Any disagreements regarding the scoring of papers were discussed by two authors (TB and CP). There is no enforced cut-off point for inclusion in a review, although Kmet et al. (2004) suggests a minimum score of 60%.

For the quantitative studies, the quality score ranged from 35% to 95%. The small sample sizes and the lack of control for confounding factors impacted on the quality rating for most studies. The scores for the qualitative studies varied widely from 42% to 100%. Most lacked a theoretical framework and/or failed to clearly describe certain aspects of the research such as aims and objectives, participant characteristics, data collection and analysis, and verification procedures for credibility. A summary of the quality scores can be found in Appendix 4. All studies contributed to the generation and analysis of themes. Despite two studies scoring 35% and 42%, it was decided that they would be included because they added to the breadth of patient experiences this review aims to cover. Any selection decisions and disagreements were resolved in consultation with a third review author (AM).

3.4.4 Data extraction and management

Data extraction from the selected studies was performed according to the CRD guidelines (University of York NHS Centre for Reviews Dissemination, 2009) and includes study setting, aims, research design, participants, type of condition and key concepts/themes relevant to the aims of this review. For the quantitative studies, the instruments for measuring psychosocial impact were also described. Extracted data of all included studies are summarised in Tables 2 and 3.

3.4.5 Data analysis and synthesis

The guidance provided by Thomas and Harden (2005 and 2008) for synthesising mixed methods evidence was used to address the review objectives. The initial parallel systematic synthesis of the evidence by method (quantitative and qualitative) followed by a third synthesis of both were instrumental in addressing such a broad review topic (Mays et al., 2005).

The first step in data analysis and synthesis was the presentation of an overview of the study characteristics with the accompanying table of extracted data (Tables 2 and 3). With the possibility of cardiac screening and/or PGT or aspects (such as the test itself, attendance at a specialist clinic or genetic counselling) considered as an exposure akin to an intervention, meta-analysis was considered for Synthesis 1 (quantitative data), however, this was not possible due to the heterogeneity of the methods and samples. Thus, a narrative approach was used (Mays et al., 2005, University of York NHS Centre for Reviews Dissemination, 2009), taking into account the differences and commonalities in the included studies in terms of participants, settings, outcome measures used and findings.

For Synthesis 2, a thematic synthesis of the qualitative studies was undertaken (Braun and Clarke, 2006, Harden and Thomas, 2005, Thomas and Harden, 2008). As illustrated in Appendix 5, the first step was to copy verbatim each paper into NVivo© software and each was read and re-read for familiarisation. Whole papers were treated as data and line-by-line coding enabled the generation of descriptive codes encapsulating the meaning of each sentence. Similarities and differences between codes were considered which allowed for arrangement within a hierarchal tree. New descriptive themes were generated to capture the meaning of clusters of initial codes. In the final step of Synthesis 2, analytical themes were then generated considering the review objectives and agreed on by three reviewers (TB, CP, and AM). This process had the advantage ensuring that the review was not constrained by any a priori frameworks (Thomas and Harden, 2008) which in turn facilitated a comprehensive view of the patient experience and allowed any appropriate frameworks to develop which is a key component of developing complex interventions (Craig et al., 2008).

Where mixed methods were conducted, the study was subdivided into the quantitative and qualitative findings. This was taken as a practical step as these were nevertheless reported in separate papers except for one paper (Hendriks et al., 2005a).

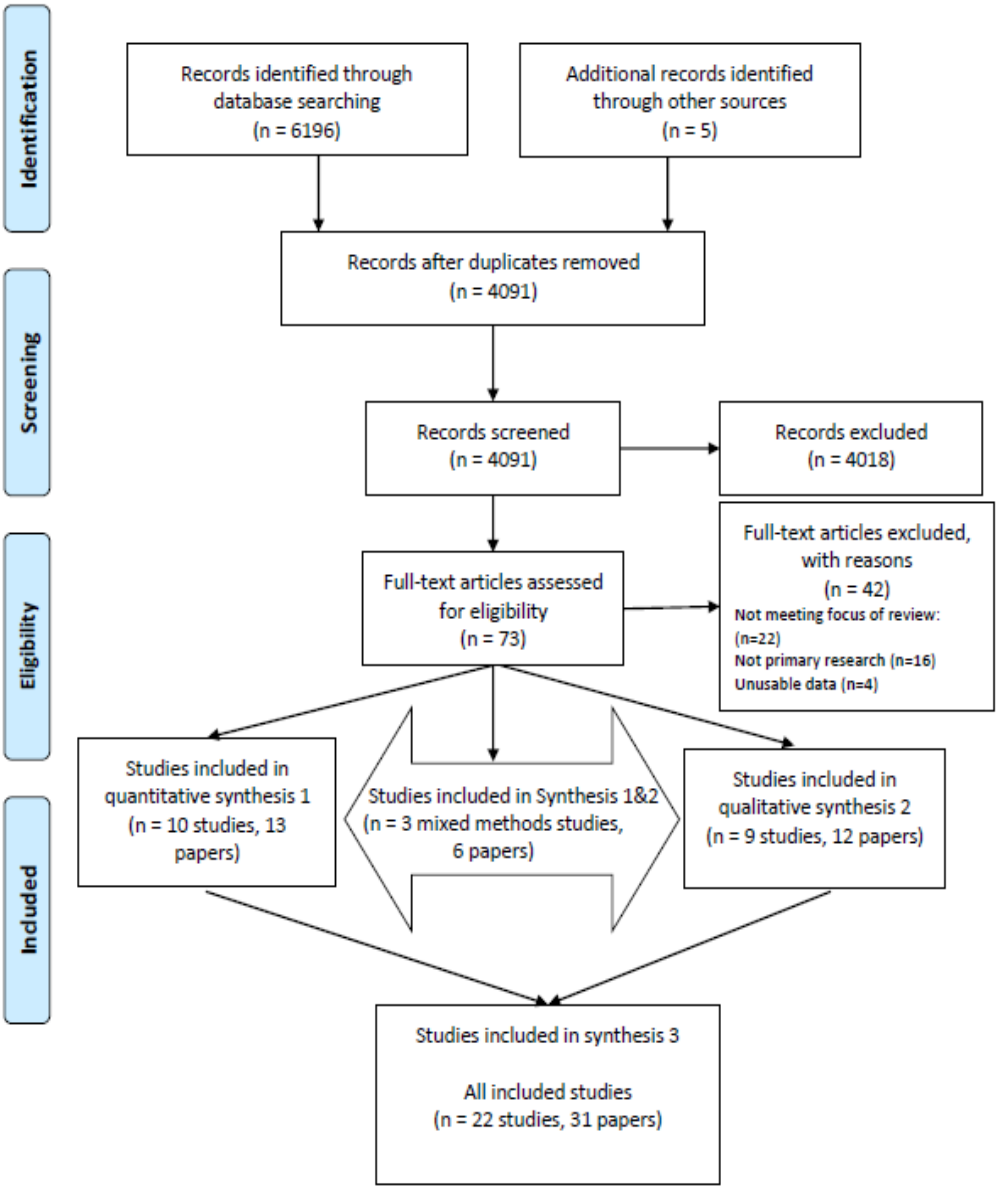
To facilitate the combination of the first two syntheses to bring about Synthesis 3, a matrix was devised adopted from Thomas and Harden (2005 and 2008) guided by the review objectives and summary findings from each synthesis. These were compared to look at consistencies and contradictions from which review conclusions and recommendations could be made.

3.5 Results

3.5.1 Overview of studies

A Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flowchart (Moher et al., 2009) detailed the methodological steps of this systematic review and provides a summary of papers included and excluded in the review (Figure 6)

Figure 6 Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) flowchart of database searches



The search results from the seven databases yielded 6201 potential citations of which 2110 were duplicates, leaving 4091 for evaluation. After reviewing the titles and abstracts, 73 papers were selected and read in full including 5 additional papers that were identified for inclusion following a search of the reference lists. Forty-two papers

were excluded for the following reasons: 22 papers not in keeping with the focus of the review in terms of population or focused solely on the yield of screening and/or PGT, 16 papers were not primary research; and four papers had unusable data (qualitative studies where quotes were not attributed according to proband or at-risk relative status).

Thirty-one papers reporting 22 studies met the inclusion criteria and although two scored below the recommended 60% cut-off for quality assessment, they were included due to the additional perspective provided to the overall review. Thirteen papers used a quantitative research design (10 studies), 12 papers employed a qualitative research design (nine studies) and a total of six papers were classified as mixed methods (three studies). Most included studies were conducted in Europe (17 studies) and there were three from North America and two were conducted in Australia.

Eligible papers were published between 2002 and June 2017 and were focused on the following ICCs: HCM, LQTS, ARVC, LQTS and SADS. In addition to an ICC, one paper included patients with Huntington disease (HD) and hereditary breast or ovarian cancer (HBOC) (MacLeod et al., 2014); and two papers included patients with familial hypercholesterolaemia (FH) (Meulenkamp et al., 2008, Smets et al., 2008). Samples included a population from the age of 5 and comprised of family members at risk for ICCs, patients affected with ICCs, partners, and parents, thus encompassing the lower limit of 16 years old and the target population of this review. Although the focus of this review were the experiences of adult patients (≥16 y/o) at risk for ICCs, it was not practicable to extract data always pertaining only to this population group within the studies where they were not exclusively represented. Therefore, the synthesis

has considered the results of the studies as a whole but where possible, emphasised the findings for this particular group.

A number of researchers used the same sample across a number of included papers and therefore classified as one study for the purposes of this review: Bratt et al. (Bratt et al., 2012, Bratt et al., 2013), Christiaans et al. (Christiaans et al., 2009a, Christiaans et al., 2009b), Etchegary et al. (Etchegary et al., 2016, Etchegary et al., 2015), Geelen et al. (Geelen et al., 2011, Geelen et al., 2012), Hamang et al. (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012), Manuel et al. (Manuel and Brunger, 2014, Manuel and Brunger, 2015) and Meulenkamp et al. and Smets et al. (Meulenkamp et al., 2008, Smets et al., 2008).

The majority of the studies focused on a population having genetic testing (20) albeit with some having had cardiac screening in the past, two studies had a sample that only focused on cardiac screening (Bratt et al., 2012, Bratt et al., 2013, McGorrian et al., 2013), and the study conducted by Christiaans et al. (Christiaans et al., 2009a, Christiaans et al., 2009b) looked at a sample where only PGT was done.

3.5.2 Synthesis 1 Quantitative studies

This synthesis of 13 quantitative studies (inclusive of the quantitative aspect of the mixed methods studies) included four prospective longitudinal studies, seven cross-sectional survey studies, one which initially conducted a cross-sectional survey then progressed to a prospective longitudinal study; and one observational study, as summarised in Table 2. The studies were conducted in Europe (ten studies), Australia (two studies) and the United States (one study). A total of 2,731 participants were included in these studies consisting of either ICC at-risk relatives only (four studies), ICC probands and at-risk relatives (eight studies); or including spouses (one study).

Six studies focused on participants impacted by HCM whilst two studies focused on LQTS; the rest were a combination of ICCs including one study which included participants with FH. Hypertrophic cardiomyopathy dominates in terms of the ICC entity as it is represented in 10 of the studies. Not all studies differentiated their findings according to these participant characteristics, so a clear breakdown of the population was not possible.

The sample sizes of the included studies varied widely; the smallest study had 28 participants (Bratt et al., 2013) and the largest, recruiting from a national registry in Finland, had 1191 participants (Hintsa et al., 2009). Across the 13 studies, the average sample size was 210 and the median sample size was 109. Most of the studies exclusively recruited adult participants, however, two studies also included participants who were below 16 years old with the lowest age for a participant reported as 5 years old. Where there were very young participants, their parents, whether they were unaffected, affected with ICCs or carriers; responded on behalf of the children.

Table 2 Characteristics of quantitative studies included in Synthesis 1

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
Bratt et al. (2012) Bratt et al. (2013) Research outpatient clinic Sweden	2013: To measure quality of life (QoL) in asymptomatic patients before and after hypertrophic cardiomyopathy (HCM) diagnosis	2013: Longitudinal prospective case-control study	2013: 13 participants diagnosed with HCM at screening compared to 15 participants with normal screens Children & adolescents aged 5-18 years old (median age 11)	2013: QoL: Lindström Model measured before diagnosis and at a median 22-month follow-up	2013: More psychosomatic symptoms in those with HCM but not at follow-up. Screening does not appear to negatively influence QoL	2013: 86% (small sample size, partial control for confounding)
Charron et al. (2002) ICC clinic France	To discuss the complex issues related to genetic testing in HCM To report preliminary experience with multidisciplinary approach	Observational study	29 participants seen for HCM (predictive genetic testing) PGT Adults aged 18-66 years old	None	No major adverse psychological effects after getting results of genetic test 10 declined genetic testing	35% (overall flaws in study design)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
Christiaans et al. (2009a) Christiaans et al. (2009b) ICC clinic Netherlands	2009a: To evaluate views on PGT counselling & testing, and follow-up in HCM mutation carriers. 2009b: To assess long-term QoL & psychological distress in those undergoing PGT & subsequent HCM mutation carriers. Identify sociodemographic, clinical, and risk and illness perception related factors that are associated with deteriorations in mutation carriers' QoL and psychological distress	Cross-sectional survey study	123 patients who had HCM PGT compared to Dutch population (total sample=228 including probands) Adults aged 30-62 years	2009a: Modified centre: developed questionnaire (Stiggelbout and Kiebert, 1997) 2009b: QoL: SF-36 Psychological distress: Hospital Anxiety & Depression Scale (HADS) Perceived risk questionnaire (Aalfs et al., 2004) Illness perception: Illness Perception Questionnaire (IPQ-R)	2009a: Genetic counselling & PGT at ICC clinic valued positively by predictively tested HCM mutation carriers, positive attitude towards cardiac follow-up, relatively low proportion receiving regular cardiac follow-up (76%). 2009b: HCM carriership does not negatively affect QoL and psychological distress, Presence of manifest HCM and HCM-related symptoms can determine impaired physical QoL. Perceptions of risk and carriership, like the perceived risk of sudden death or belief in serious consequences of carriership, were the main determinants of QoL and psychological distress.	2009a: 91% (study instrument not validated, partial control for confounding) 2009b: 95% (partial control for confounding)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
Hamang et al. (2010) Hamang et al. (2011) Hamang et al. (2012) 3 Medical genetic departments Norway	To investigate the health status of patients at risk of inherited arrhythmia prior to the genetic counselling session To investigate the role of heart-focused anxiety in relation to general anxiety, depression and physical health in patients referred to specialised ICC clinics for genetic investigation and counselling To compare symptoms of heart-focused anxiety in patients with a clinical diagnosis of Long QT Syndrome (LQTS) or HCM and in patients at genetic risk of LQTS or HCM To investigate the independent influence on heart-focused anxiety of the following factors: sudden cardiac death in close relatives; a recent cardiac death of a relative; patient knowing whether other relatives previously have undergone genetic testing	2010 & 2011 Cross-sectional survey study 2012 Longitudinal prospective study	2010: 95 patients referred for predictive genetic testing compared to Norwegian population (total sample=127 including probands) 2011 & 2012: 94 patients referred for predictive genetic testing compared to Norwegian population (total population=126 including probands) Adults	2010 & 2011: QoL: SF-36 Psychological distress: HADS Heart-focused anxiety: Cardiac Anxiety Questionnaire (CAQ) Measured before genetic counselling (GC) 2012: In addition to above: Self-efficacy expectations: Bergen Genetic Counselling Self-Efficacy Scale (BGCSES) (2 weeks before GC) Satisfaction with GC: Satisfaction with Genetic Counselling Scale (SGCS) (straight after GC) Heart focused anxiety: CAQ (2 weeks before GC, 4 weeks, 6 months & 1 year after GC)	For whole group, living with genetic risk of arrhythmia and possible sudden death is most likely related to health status vulnerability, perception of current health; health outlook and resistance to illness is lower than the general population. Those who came for PGT had better QoL but had higher levels of anxiety compared to the general population. General anxiety and depression levels seemed to be unrelated to a clinical diagnosis but to living with a genetic risk of a life-threatening disorder, uncertainty regarding cardiac symptoms and high levels of heart-focused anxiety. Heart-focused anxiety was overall higher in patients with LQTS or HCM compared to at risk patients, but this did not have an independent effect in predicting heart-focused anxiety over time; a family history of sudden cardiac death in close relatives and uncertainty whether other relatives had undergone genetic testing seemed to be predisposed to heart-focused anxiety; satisfaction with the procedural parts of genetic counselling was predictive of decreased levels of heart focused anxiety	2010 & 2011 86% (small sample size, no control for confounding) 2012 82% (small sample size, no control for confounding, high number of dropouts)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
Hendriks et al. (2005) Hendriks et al. (2008) ICC clinic Netherlands	To investigate the extent and course of distress caused by predictive genetic testing in LQTS	2005: Mixed methods 2008 Prospective longitudinal survey study	2005 7 members of LQTS family who underwent screening (spouses n=4) 2008 77 patients undergoing cardiac investigation then PGT for LQTS and their partners (n=57) Adult patients	2005 Anxiety: Spielberger State Anxiety Inventory Depression: Beck's Depression Inventory (BDI) Disease impact: Impact of Events Scale (IES) Measured at first clinic attendance, after test results and at 18 months. 2008 Disease-related anxiety: IES Depression: BDI Measured within 2 weeks of first visit, 2 weeks after first timepoint & 18 months after receiving results.	High distress scores even in those who are not affected. Cardiac investigation followed by predictive genetic testing in LQTS leads to distress but return to normal levels within 18 months. Carriers with an uncertain electrocardiogram (ECG) had a higher incidence of clinical distress most likely due to heralding the disease.	2005 42% (no theoretical framework, data collection methods unclear, no description of data analysis, verification procedures and reflexivity) 2008 86% (small sample size, no control for confounding)
Hintsa et al. (2009) University Hospital Finland	To examine whether depressive symptoms are related to arrhythmic events among symptomatic and asymptomatic LQTS mutation carriers and syncope events among their relatives not carrying the family's LQTS-causing mutation.	Cross-sectional survey study	1191 patients who have had genetic testing for LQTS from the Finnish LQTS registry, carriers (n=569) were compared to non-carriers (n=622) 16-65 years	Depression: BDI	Depressive symptoms were associated with arrhythmic events but not with being a carrier of LQTS.	91% (no control for confounding)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
Hoedemaekers et al. (2007) ICC clinic Netherlands	To investigate the influence of coping styles and perceived control on emotional distress in persons at risk for inherited cardiac conditions (ICCs)	Prospective longitudinal survey study	108 patients at risk for ICC attending for predictive testing ≥18 years	Coping styles: Mastery-Pearlin's mastery list Disease-specific control-Health Locus of Control (HLOC)- Coping- Threatening Medical Situations Inventory (TMSI) Emotional distress: General Health Questionnaire (GHQ)-12 Measured after first consultation, after receiving results and 2 months after	No difference in emotional distress between participants and the average Dutch citizen before or after test results. Higher degree of perceived control buffers against the onset of emotional distress.	86% (small sample size, no control for confounding, high drop-out rate for last 2 data collection timepoints)
Ingles et al. (2008) ICC clinic Australia	To identify the psychosocial factors that impact on the emotional well-being of those attending a specialty ICC clinic	Cross-sectional survey study	25 at-risk relatives for HCM attending the clinic (total population=109 including probands) ≥17 years	Psychological distress: HADS Adjustment to HCM: Patient experience scale Satisfaction with staff: patient relationship-Patient satisfaction scales	Nearly all participants showed high to very high satisfaction with the clinical relationship developed with staff 24% with HCM say they have adjusted to diagnosis but only 10% with HCM and 4% of at-risk relatives have low worry scores	86% (small sample size, no control for confounding)
Ingles et al. (2012) ICC clinic Australia	To identify whether there were changes in Health-Related Quality of Life (HR-QoL) following genetic testing for an ICC in patients with clinical disease and asymptomatic family members.	Prospective longitudinal survey study	21 at-risk relatives for ICC attending the clinic (total population=54 including probands) compared to general Australian population	QoL: SF-36 Measured before genetic results were given, then 1-3 months, 6 and 12 months after the result was given	Both affected and at-risk relatives had no change in HR-QoL No significant difference between groups	86% (small sample size, no control for confounding)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
			>15 years			
Jensen et al. (2013) ICC clinic Denmark	To study the outcome of clinical screening and predictive genetic testing of child relatives from HCM families and assessed the age-related penetrance of HCM during 12 years of follow-up	Cross-sectional survey study for psychological evaluation aspect of the study	38 patients (12 carriers, 26 at-risk relatives who are untested or no genetic testing available for HCM. (Total population=41 including probands) compared to 23 non-carriers <18 years at recruitment but majority >18 at 12-year f/u	Psychological distress: HADS, IES and State Trait Anxiety Inventory Negative affectivity and social inhibition: DS14 Prevalence of Type D personality	No significant differences in anxiety, depression, type D personality, or overall psychological impact of participation in the family screening program between the 3 groups	77% (study design for psychological study not well described, small sample size, no control for confounding)
Khouzam et al. (2015) National support group HCM Association United States	To assess factors associated with the underutilisation of genetic services for HCM	Cross-sectional survey study	36 at-risk for HCM, (total population=306 including probands) >18 years	Factors associated with an individual's decision to obtain genetic testing & counselling for HCM based on the Health Belief Model (HBM) (Cyr et al., 2010)	HBM components of cues to action (that genetic testing had been discussed or offered) and perceived benefits (that genetic testing can help family members and make better family healthcare decisions) and barriers had greatest impact on genetic testing	86% (small sample size, no control for confounding)

Study Setting/ Country	Aim	Design	Participants	Psychosocial impact tool/Satisfaction questionnaire	Main findings	Kmet (2004) score and quality issues
McGorrian et al. (2013) ICC clinic Republic of Ireland	To define the anxiety and depression burden associated cardiac screening, whether these traits cluster within families, and to examine the associates of higher levels of anxiety and depression states in this population	Cross-sectional survey study	316 patients at-risk relatives for inherited CM or channelopathy, Sudden Adult Death Syndrome (SADS) or Sudden Infant Death Syndrome (SIDS) ≥16 years	QoL: SF-12 Psychological distress: HADS Measured before cardiac screening	Overall high levels of anxiety Younger patients also had high rates of clinically significant anxiety Higher levels of anxiety and depression tends to run in families	86% (28% of sample second degree relatives who may not have risk, no control for confounding)
Meulenkamp et al. (2008) Smets et al. (2008) ICC clinic & Lipid clinic Netherlands	Smets et al. (2008): To explore how QoL of carriers compare to the Dutch general population; and to what extent the carrier's QoL and their parents' perception concur	Smets et al. (2008): Cross-sectional survey study	Smets et al. (2008): 35 mutation carriers for HCM, LQTS & Familial Hypercholesterolaemia (FH) who had genetic testing at least half a year ago compared to Dutch reference group and their parents' responses 8-18 years (23 patients aged 12-18 years)	Smets et al. (2008): QoL-KIDSCREEN	Smets et al. (2008): No statistically significant differences in scores between carriers and the reference group No differences were found between carriers and their parents' ratings, but parents rated their child's psychological well-being significantly lower	Smets et al. (2008): 86% (small sample size, no control for confounding)

3.5.2.1 Quality of life in at-risk family members undergoing cardiac screening and/or predictive genetic testing for inherited cardiac conditions

The quantitative studies reporting on quality of life for (QoL) at-risk patients undergoing PGT concluded that there was no significant difference in QoL compared to the general population (Christiaans et al., 2009b, Christiaans et al., 2009a, Smets et al., 2008) and this continues to be observed over time in the longitudinal studies (Bratt et al., 2013, Ingles et al., 2012). One study reported that those who underwent PGT had better QoL than the general population (Hamang et al., 2010, Hamang et al., 2012). However, once signs and symptoms appear or an ICC diagnosis is made, the QoL appears to be worse compared to the general population (Christiaans et al., 2009a, Christiaans et al., 2009b, Hamang et al., 2010, Hamang et al., 2012). This trend was not demonstrated in Ingles et al. (2012) even if patients had symptoms after one year of follow up. Furthermore, two studies which also compared QoL in asymptomatic carriers and those affected by an ICC did not see any significant difference between both groups (Ingles et al., 2012, Jensen et al., 2011).

McGorrian et al. (2013) reported that older participants (≥55 y/o) had lower scores for the physical aspect of the SF-12, indicating worsening physical health, but did not attribute this to the presence of signs and symptoms specific to ICCs. Hamang et al. (2010, 2011, 2012) saw a similar relationship to increasing age and decreased physical functioning based on the SF-36 scores.

3.5.2.2 Psychological distress and depression in at-risk family members undergoing cardiac screening and/or predictive genetic testing for inherited cardiac conditions

There was conflicting evidence regarding anxiety and psychological distress in participants who had prior cardiac screening followed by PGT compared to the general

population: one study reported no difference (Hoedemaekers et al., 2007); two studies demonstrated that the at-risk for ICC group was more anxious (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012, Hendriks et al., 2005a, Hendriks et al., 2008); and Christiaans et al. (2009a & 2009b) reported that the ICC group was less anxious. Overall, there was an initial stage of high anxiety, shock, and distress when participants received their PGT results (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012, Hendriks et al., 2005a, Hendriks et al., 2008). Although Hoedemaekers et al. (2007) reported no difference in psychological distress for those at risk for ICCs and the general population, nevertheless there were between 16.37%-23.2% of the participants who had clinical levels of distress at the three timepoints.

There were also high levels of psychological distress in patients undergoing screening. Ingles et al. (2008) reported low worry scores in only 1 out of the 10 at-risk family members whilst McGorrian et al. (2013) found that 19.2% of their patients had high psychological distress.

Overall, depression does not feature prominently in at-risk patients undergoing screening and/or PGT for ICCs (Charron et al., 2002, Ingles et al., 2008, Jensen et al., 2011), equalling (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012, Hendriks et al., 2005a, Hendriks et al., 2008) or even having better scores (Christiaans et al., 2009a, Christiaans et al., 2009b) than the general population. However, in a population of LQTS carriers, depressive symptoms were prevalent when accompanied by arrhythmic events (Hintsa et al., 2009). Anxiety and depression scores were also found to vary between families rather than within families, indicating a greater predisposition for emotional distress in certain families (McGorrian et al., 2013).

Two studies discussed a transition and adjustment to living with the results of screening and/or PGT. This is evident in the shift to normalisation of anxiety in most patients as

time goes by (Hendriks et al., 2005a, Hendriks et al., 2008) and a decrease in psychosomatic symptoms after a diagnosis of HCM at follow-up (Bratt et al., 2013).

3.5.2.3 Factors impacting quality of life, depression, and psychological distress in those at-risk for inherited cardiac conditions

Three studies reported that a clinical diagnosis and/or signs and symptoms of an ICC, rather than carriership for a pathogenic ICC variant, contributes to lower QoL and depression (Christiaans et al., 2009a, Christiaans et al., 2009b, Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012, Hintsa et al., 2009). Furthermore, Christiaans et al. (2009 a & b) and Hamang et al. (2010, 2011 & 2012) concur that perceptions of the possibility of sudden cardiac death have a major impact on QoL and psychological distress.

High anxiety appears to be associated with uncertainty of the cause of cardiac-related symptoms (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012) or diagnostic uncertainty (no definitive cardiac screening results) (Hendriks et al., 2005a, Hendriks et al., 2008). Other predictors for a high anxiety score were family-related: a close relationship with the proband, other family members with high anxiety, not having a partner (McGorrian et al., 2013); and worry about other family members not having PGT (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012). McGorrian et al. (2013) also cited that a lower educational level was related to higher anxiety scores whilst Hamang et al (2010, 2011, 2012) found that a higher educational level was associated with better outcomes in the five out of the eight SF-36 health status domains.

3.5.2.4 Experience with inherited cardiac conditions clinical services

Most of the studies recruited participants representing the patients in emerging specialist ICC clinics and therefore also evaluated their clinical services. Participants had a high satisfaction with the relationship they had with their clinicians (Ingles et al., 2008). Indeed, satisfaction with genetic counselling appeared to decrease heart–focused

anxiety and promoted adjustment to a diagnosis (Hamang et al., 2010, Hamang et al., 2011, Hamang et al., 2012). When genetic testing was offered and was perceived to be useful for the participant and their family, participants will still tend to take up testing despite possible negative consequences (impact on insurance) (Charron et al., 2002).

However, Christiaans et al. (2009 a & b) reported that whilst participants valued the services of the ICC clinic, there was a relatively low proportion of patients attending clinic. The earliest study in this review reported that despite the multi-disciplinary approach of their clinic, ten of the 29 participants did not engage in PGT and said they 'would rather not know' and felt that they already had the pathogenic variant although it was not clear if they had signs and symptoms (Charron et al., 2002).

3.5.3 Synthesis 2 Qualitative studies

This synthesis of 12 qualitative studies (inclusive of the qualitative aspect of the mixed methods studies), as summarised in Table 3, included ten studies conducted in Europe (four from the Netherlands) and two from North America, specifically Canada. All studies were conducted using interviews which were described as semi-structured or in-depth and in two studies, whilst most participants were interviewed individually, some were interviewed in pairs (van der Werf et al., 2014) or in focus groups (Manuel and Brunger, 2014, Manuel and Brunger, 2015). Three studies conducted interviews at more than one timepoint (Geelen et al., 2012, Geelen et al., 2011, Hendriks et al., 2008, Hendriks et al., 2005a, Manuel and Brunger, 2014, Manuel and Brunger, 2015). The participants included in these studies consisted of either ICC at-risk relatives only (five studies), ICC probands and at-risk relatives (two studies); or including spouses and partners (four studies); and one study included all of these alongside parents.

For each of these ICCs, there were two studies each focused solely on the following individual conditions: HCM (Bratt et al., 2013, Bratt et al., 2012, Geelen et al., 2012, Geelen et al., 2011), LQTS (Andersen et al., 2008, Hendriks et al., 2008, Hendriks et al.,

2005a) and ARVC (Etchegary et al., 2016, Etchegary et al., 2015, Manuel and Brunger, 2014, Manuel and Brunger, 2015). Four studies were a combination of participants impacted by HCM and LQTS (Ormondroyd et al., 2014, Smart, 2010, Whyte et al., 2016), including one study which included those with Familial Hypercholesterolemia (FH) (Meulenkamp et al., 2008, Smets et al., 2008). One study focused on at-risk relatives undergoing screening for SADS (van der Werf et al., 2014) and one study included HCM at-risk relatives alongside those with HD and HBOC (MacLeod et al., 2014). Hypertrophic cardiomyopathy and LQTS dominate in terms of the ICC entity as they are represented individually or in combination in nine of the studies. Nine studies exclusively recruited adult participants although amongst these, four studies did not report the age range of their participants. The remaining three studies also included participants who were below 16 years old with the lowest age for a participant reported as 8 years old; but these children were all individually interviewed.

All the studies included attributed quotes according to these participant characteristics, which made it possible to code quotes given by the at-risk family members who are the focus of this review to facilitate thematic synthesis. The emergent codes, descriptive themes and analytical themes are presented in Figure 7.

Table 3 Characteristic of Qualitative Studies Included in Synthesis 2

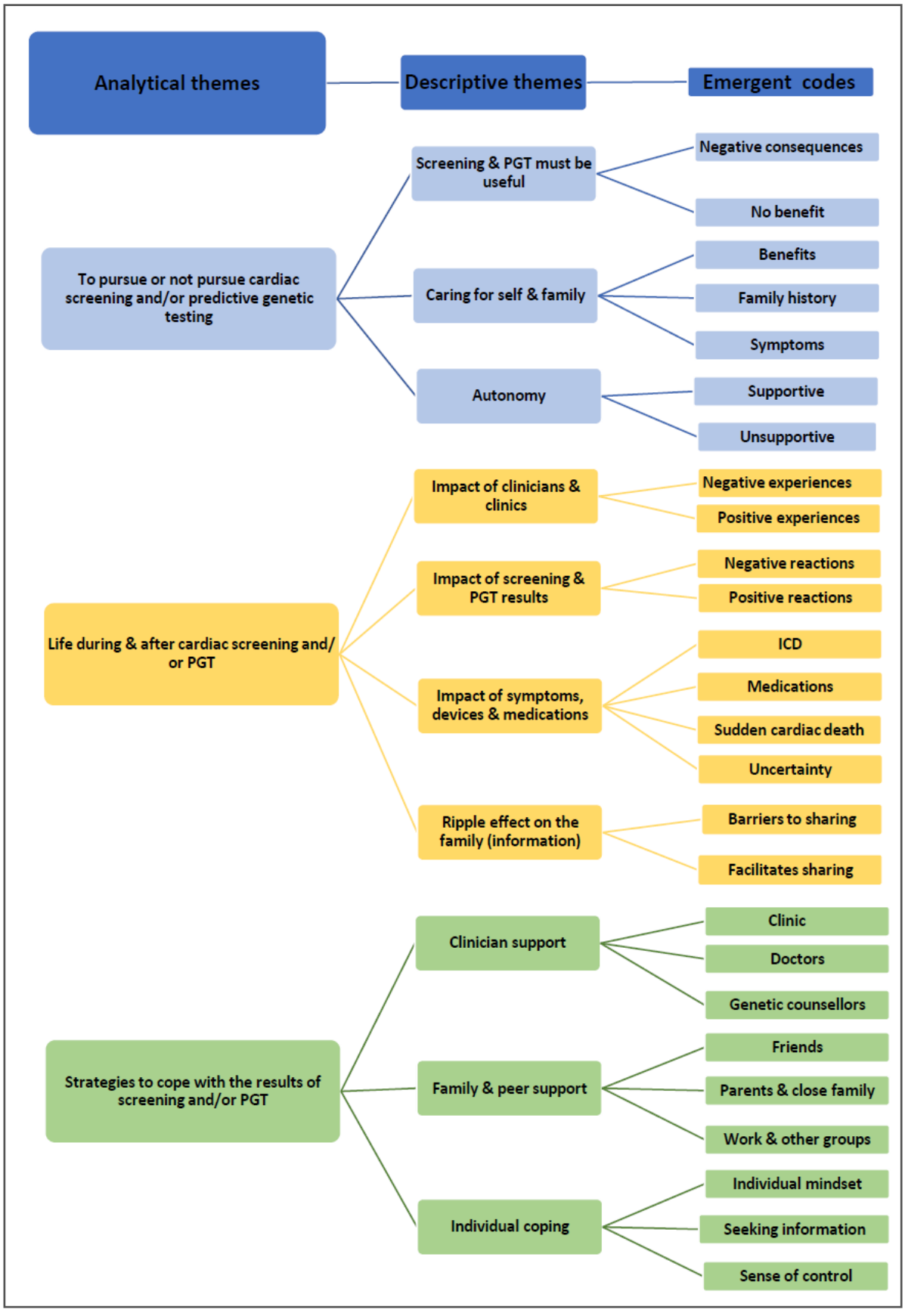
Study Country/ Setting	Aim	Design	Participants	Main findings Descriptive themes	Kmet (2004) score and quality issues
Andersen et al. (2008) University hospital Norway	To investigate psychosocial aspects of living with Long QT Syndrome (LQTS) & experiences with healthcare services	In-depth interviews Systematic Text Condensation (Giorgi, 1985)	4 patients who had predictive genetic testing (PGT) for LQTS 23-76 years	Early and gradually acquired knowledge of syndrome is an advantage. Main concern is for children/grandchildren. Minimal knowledge amongst healthcare professional about LQTS	90% (no theoretical framework or comment on reflexivity)
Bratt et al. (2012) Bratt et al. (2013) Research outpatient clinic Sweden	2012: To describe the experiences of patients who screened positive for Hypertrophic Cardiomyopathy (HCM) and its impacts on their daily life.	2012: In-depth interviews. Qualitative content analysis (Graneheim and Lundman, 2004)	2012: 13 participants diagnosed with HCM at screening 8-18 years	2012: Involuntary change experienced affecting daily life with limitations both on an individual and social context. Reorientation brings hope and faith in the future.	2012: 90% (no theoretical framework or comment on reflexivity)
Etchegary et al. (2015) Etchegary et al. (2016) ICC clinic Canada	To explore the perspectives of individuals who undergo genetic testing to inform the provision of health services and promote informed decision making	Semi-structured interviews. Qualitative description (Sandelowski, 2000)	21 patients at-risk, carrier, non-carrier or spouse of patient with TMEM43 p.S358L mutation for Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) (Total population=21 including spouses) >13 years (mean age 44 years)	Strong need to rule our risk for family rather than for themselves. Lingering doubt despite a negative test. Interdependent nature of genetic test decisions. Perceived economic burden of ARVC range from employment, career choices, insurance worries, decreased household spending and the need for childhood employment.	90% (no theoretical framework or comment on reflexivity) 90% (no theoretical framework or comment on reflexivity)

Study Country/ Setting	Aim	Design	Participants	Main findings Descriptive themes		Kmet (2004) score and quality issues
Geelen et al (2011) Geelen et al (2012) ICC Clinic Netherlands	To contribute to the discussion on genetic testing of children, from the perspective of the concrete, everyday lives of 'HCM families'. How the views on genetic testing in 'the child's best interest' are constructed To have a better understanding of the origins and backgrounds of fears of genetic discrimination and its possible impact on the uptake of HCM genetic testing	Longitudinal semi-structured interviews over 3.5 years. Thematic analysis.	57 participants from 6 extended families dealing with HCM (affected, mutation carriers and non-carriers, at-risk members, spouses, parents), unsure of status of each, quotes identified status of individual Ages not stated	The best interest of the child is considered in the context of the family. This best interest can change over time as families engage in their own way with genetic testing as science progresses. Pre-existing experiences of discrimination influences uptake of genetic testing		90% (no theoretical framework or comment on reflexivity) 90% (no theoretical framework or comment on reflexivity)
Hendriks et al. (2005) Hendriks et al. (2008) ICC clinic Netherlands	To investigate the extent and course of distress caused by predictive genetic testing in LQTS	2005: Mixed methods 2008 Prospective longitudinal survey study	2005 7 members of LQTS family who underwent screening (spouses n=4) 2008 77 patients undergoing cardiac investigation then PGT for LQTS and their partners (n=57) Adult patients	2005 Anxiety: Spielberger State Anxiety Inventory Depression: Beck's Depression Inventory (BDI) Disease impact: Impact of Events Scale (IES) Measured at first clinic attendance, after test results and at 18 months. 2008 Disease-related anxiety: IES Depression: BDI Measured within 2 weeks of first visit, 2 weeks after first timepoint & 18 months after receiving results.	High distress scores even in those who are not affected. Cardiac investigation followed by predictive genetic testing in LQTS leads to distress but return to normal levels within 18 months. Carriers with an uncertain ECG had a higher incidence of clinical distress most likely due to heralding the disease.	2005 42% (no theoretical framework, data collection methods unclear, no description of data analysis, verification procedures and reflexivity) 2008 86% (small sample size, no control for confounding)

Study Country/ Setting	Aim	Design	Participants	Main findings Descriptive themes	Kmet (2004) score and quality issues
MacLeod et al. (2014) Genetics clinic United Kingdom	To look at motivation of participants to be tested when young, their experiences of the counselling process and the advice they would offer to health professionals and other young adults considering testing.	Telephone interviews. Interpretative phenomenological analysis	10 patients who had a predictive test for HCM (total population=36 with other conditions included) 15-25 years	No regrets with being tested. Value of genetic counselling is for information/support and not for facilitating a decision. In HCM, parents were a strong influence for genetic testing.	90% (no theoretical framework or comment on reflexivity)
Manuel & Brunger (2014) Manuel & Brunger (2015) Genetics clinic Canada	To describe the experience of predictive genetic testing for ARVC in the context of novel gene discovery. To understand how individuals living in a family at risk for ARVC make health care decisions in the context of risk perception.	9 Individual interviews, 3 focus groups, 5 follow up interviews. Grounded theory (Glaser et al., 1968)	21 patients involved in decision making for ARVC PGT (total population=29 including spouses)	Decision-making for predictive genetic testing develops gradually over time or happens so quickly that it is a *'fait accompli'.* Key factors identified by the family: scientific process, deaths in the family, signs of disease, gender, relational responsibility, and family support. Families living with ARVC juxtapose existing scientific knowledge with experiential knowledge in the process of 'awakening' to concept of risk.	95% (no comment on reflexivity) 95% (no comment on reflexivity)
Meulenkamp et al. (2008) Smets et al. (2008) ICC clinic Lipid clinic Netherlands	Meulenkamp et al. (2008): To articulate the experiences of mutation carriers as to the way they perceive their carrier status and their experiences concerning lifestyle modifications, medication-use and worries to provide insight into coping.	Meulenkamp et al. (2008): Interviews Leventhal's model of self-regulation (Cameron and Leventhal, 2003)	Meulenkamp et al. (2008): 17 HCM & LQTS mutation carriers found through predictive genetic testing (total population with Familial Hypercholesterolaemia (FH) patients=33) 8-18 years (23 patients aged 12-18 years)	Participants expressed positivity but feelings of control varied. Issues with adherence and side-effects expressed. They coped with worries of dying and difference from peers by expressing faith in medications, trying to be similar to peers or in contrast, be 'different'.	95% (no comment on reflexivity)

Study Country/ Setting	Aim	Design	Participants	Main findings Descriptive themes	Kmet (2004) score and quality issues
Ormondroyd et al. (2014) ICC clinic United Kingdom	To explore the process of PGT within families: to understand how people learn about risk and make decisions about undergoing testing. To evaluate the psychosocial impact of testing, explore attitudes to direct contact of relatives about the ICC and availability of testing.	Interviews. Thematic analysis.	22 patients who have undergone PGT for HCM & LQTS ≥18 years	Concept of risk and meaning of being a carrier without manifest disease can be a factor in dissemination to family members. Testing is pursued to rule out risk for self and children. There are concerns for testing of children at a young age, but young participants are pragmatic about their results.	90% (no theoretical framework or comment on reflexivity)
Smart (2010) ICC clinic United Kingdom	To look at patient experiences of genetic testing and cascade screening for HCM and LQTS focusing on potential impediments to testing and screening.	Semi-structured interviews Analytical hierarchy	9 patients who have undergone PGT for HCM & LQTS (total population=27 including probands) Ages not stated	Testing is a way to provide health information for self and children. Ambivalence about value and impact of testing. Concerns raised about communicating risk for family.	90% (no theoretical framework or comment on reflexivity)
van der Werf et al. (2014) ICC clinic Netherlands	To study the experiences and attitudes of first-degree relatives who attended an ICC clinic for evaluation for a family history of Sudden Adult Death Syndrome (SADS)	In-depth interviews Inductive	9 adult relatives of young victims of SADS (total population=10 including 1 spouse) Ages not stated	Medical professionals had minimal role in facilitating screening. Mourning process hampered search for information and main reason for attending clinic is the need to understand cause of SADS and to prevent recurrence.	90% (no theoretical framework or comment on reflexivity)
Whyte et al. (2016) Genetics clinic Republic of Ireland	To gain a better understanding of the process of family communication from individuals who had received either a positive or a negative predictive test result for an inherited cardiac condition (ICC)	Semi-structured interviews Thematic analysis (Braun and Clarke, 2006)	9 patients who had PGT for HCM & LQTS Ages not stated	Knowledge of genetic information has a positive effect on families. Future generations, gender, proximity, and lack of contact play a part in family communication.	90% (no theoretical framework or comment on reflexivity)

Figure 7. Synthesis 2 emergent codes, descriptive themes, and analytical themes



Abbreviations: ICD-Internal Cardioverter Defibrillator, PGT-Predictive Genetic Test

3.5.3.1 Analytical theme 1: To pursue or not pursue cardiac screening and/or predictive genetic testing

Cardiac screening and/or PGT must be useful

Across the studies, participants discussed what led them to make the decision to pursue cardiac screening and/or PGT and the barriers and facilitators to this were numerous and inter-related. A major barrier and source of ambivalence for pursuing screening and/or PGT was the perception that this would be of no benefit and may even be a disadvantage to know their carrier status for an ICC, providing little information on prognosis yet may result in being labelled with a condition considered a burden in everyday life (Smart, 2010). They could also experience 'genetic discrimination' with the accompanying problems with employment and insurance (Geelen et al., 2011, Geelen et al., 2012). Furthermore, participants questioned the purpose of knowing when definitive treatments may not available (particularly for HCM) (Geelen et al., 2011, Geelen et al., 2012, van der Werf et al., 2014) and in this context described the screening process as 'over-the-top' (van der Werf et al., 2014).

Caring for self and family

First amongst the facilitators for undergoing cardiac screening and/or PGT was if participants perceived benefits could be gained from the process. On an individual level, a factor for some in seeking screening and/or testing was gaining knowledge of personal risk due to the inherited nature of the cardiac disease (Etchegary et al., 2015, Etchegary et al., 2016, MacLeod et al., 2014, Manuel and Brunger, 2014, Manuel and Brunger, 2015, Ormondroyd et al., 2014). Two studies described participants who decided to engage in screening and/or testing because it was information they just 'had to have' and 'need to know' personally (Andersen et al., 2008, Etchegary et al., 2015). They did not elaborate on their decision-making process.

The perceived benefits to the family (or for planning one) overrode the personal reasons for screening and/or PGT as many at-risk parents pursued screening and/or testing to

pave the way for testing their children and grandchildren (Andersen et al., 2008, Bratt et al., 2012, Bratt et al., 2013, Etchegary et al., 2015, Etchegary et al., 2016, Geelen et al., 2011, Geelen et al., 2012, MacLeod et al., 2014, Manuel and Brunger, 2014, Manuel and Brunger, 2015, Meulenkamp et al., 2008, Ormondroyd et al., 2014, Smart, 2010, Smets et al., 2008, van der Werf et al., 2014, Whyte et al., 2016). It was a way of ensuring that they have done all they can to access early treatment and preserve the health of their child. This caretaking response was extended to other family members in that the oldest sibling starts the process so they can 'drag everyone else along' (Etchegary et al., 2015, Etchegary et al., 2016) and a sense of relational responsibility to negotiate a course of action with a spouse (Etchegary et al., 2015, Etchegary et al., 2016, Manuel and Brunger, 2014, Manuel and Brunger, 2015). Consideration for the risk for future generations, even in participants in their late teens (Bratt et al., 2012, Bratt et al., 2013, MacLeod et al., 2014) was already apparent, leading to conflicting thoughts for those who were already parents that having another child who is potentially affected could be a burden whilst at the same time, a pregnancy at-risk for an ICC would not be grounds for a termination (Andersen et al., 2008).

Another motivator to engage in screening and/or testing is the family history of the individual. In communities where the ICC is due to founder mutations (Etchegary et al., 2015, Etchegary et al., 2016), or have lived for a long time with the existence of a genetic condition in their family (Manuel and Brunger, 2014, Manuel and Brunger, 2015), the process of genetic testing was sometimes thought of as a non-event and was not a difficult or stressful decision to make. Participants felt that they were contributing to the advancement of scientific knowledge by having a genetic test (Manuel and Brunger, 2014, Manuel and Brunger, 2015).

If there have been young, sudden deaths due to an ICC in the family, undergoing investigations gave participants peace of mind and considered this as part of the mourning process for their loved-one (van der Werf et al., 2014). Some participants

decided it was time to pursue this when a pattern of illness and death emerged, they are at the same age of the relative who died, or they start having similar signs and symptoms (Etchegary et al., 2016, Etchegary et al., 2015, Manuel and Brunger, 2014, Manuel and Brunger, 2015). They felt that they could take the necessary course of action to prevent these consequences (MacLeod et al., 2014, Ormondroyd et al., 2014, Whyte et al., 2016). However, having this tragic family history also made some participants feel that they had no choice but to be evaluated or tested, that 'sticking their head in the sand' not an option (Geelen et al., 2011, Geelen et al., 2012, Manuel and Brunger, 2014, Manuel and Brunger, 2015, van der Werf et al., 2014).

Autonomy

The studies showed autonomy in decision making may be at risk as participants expressed the concern for family often overrode the concern for personal health and many stated that they might not have pursued testing if it were not for their children (Etchegary et al., 2015, Etchegary et al., 2016, Geelen et al., 2011, Geelen et al., 2012). Parents do not fear want happens to them, only for the welfare of their child should they lose a parent due to an ICC (Andersen et al., 2008).

For many participants, particularly those in their teens, engaging in screening and/or testing was imposed on them by parents (Etchegary et al., 2015, Manuel and Brunger, 2014, Ormondroyd et al., 2014). Participants in other studies who also reported pressure from family to undergo testing and/or screening initially mentioned a feeling of resentment but admitted that they felt their relatives had their best interest at heart (MacLeod et al., 2014, Smart, 2010). However, in one study, some voiced that they felt they were not free to make their own decision and expressed resentment (Manuel and Brunger, 2014, Manuel and Brunger, 2015).

This potential conflict of preserving autonomy versus pressure to pursue screening and/or PGT for family considerations was illustrated in the opinions of participants about

other relatives who may be at-risk. It was strongly stated that relatives should be informed and that they should take up screening or testing. This was thought to be a positive action and the relatives' right not to know was not as important as their responsibility to their families (Etchegary et al., 2015). Relatives declining screening and/or testing were viewed as irresponsible and selfish (Smart, 2010, Whyte et al., 2016).

3.5.3.2 Analytical theme 2: Life during and after cardiac screening and/or predictive genetic testing

Impact of clinicians and clinics

In the earlier studies and studies that dealt with experiences of families since the early 1980s, participants did not have a favourable view of their clinicians. Health care providers were described as incompetent, particularly when participants received confusing information, incorrect treatments and medications, their reports of symptoms not acknowledged or believed; and when deaths continued despite the knowledge of an ICC in the family (Andersen et al., 2008, Manuel and Brunger, 2014, Manuel and Brunger, 2015).

In contrast, attendance in a specialist cardiac genetics clinic and undergoing genetic counselling was viewed as a positive experience by the participants (Ormondroyd et al., 2014), although for some, it was just a means to an end (MacLeod et al., 2014).

Participants had contrasting views on the speed at which they were seen in a specialist clinic; others felt that there was hardly any wait whilst some felt it was not quick enough (van der Werf et al., 2014). This delay affected those who attended non-specialised clinics even more as there was a long wait between multiple appointments (MacLeod et al., 2014, van der Werf et al., 2014).

Overall, genetic counsellors were described as warm and supportive (van der Werf et al., 2014) and information was delivered in a manner that was understood by the patient

(Andersen et al., 2008, MacLeod et al., 2014). More tailoring of counselling, especially for young people, was mentioned as an area for improvement as there were instances where PGT was explicitly presented by healthcare professionals simply as ‘a good thing’ (MacLeod et al., 2014). Participants also suggested more direct guidance regarding screening from a medical professional, particularly in SADS families because their thoughts and emotions are focused on the mourning process (van der Werf et al., 2014).

Impact of screening and/or genetic testing results

The qualitative studies gave more insight into the specific psychosocial impact that the screening and/or genetic testing process and results can have on patients. Getting health information through screening and/or genetic testing was considered a helpful and positive process, regardless of result (MacLeod et al., 2014, Ormondroyd et al., 2014, Whyte et al., 2016). There was a practical aspect to the information from the test such as helping patients adjust their lifestyle, plan for their children and reproductive options (Andersen et al., 2008).

Nevertheless, the process can also bring about strong emotions. Those with a family history of sudden death were highly distressed when they were screened/tested soon after the tragic event and had a very close relationship with the deceased (Meulenkamp et al., 2008, Smets et al., 2008). Furthermore, a study on SADS screening mentioned how the process brought back painful memories (van der Werf et al., 2014).

Waiting for screening or PGT results has been described as a ‘very dark period’ (Ormondroyd et al., 2014). Participants also worried about the consequences of a carrier result for a number of things such as changes in their own and their children’s insurance policies (Geelen et al., 2012, Geelen et al., 2011), mortgages, loss of livelihood and lifestyle changes (Manuel and Brunger, 2014, Manuel and Brunger, 2015, van der Werf et al., 2014); and even adverse effects on marriageability (Ormondroyd et al., 2014).

Overall, there was an initial stage of high anxiety, shock, and distress when participants received results confirming an ICC diagnosis or genetic carrier status (Hendriks et al., 2005a, Hendriks et al., 2008, MacLeod et al., 2014, Ormondroyd et al., 2014, Smart, 2010). This applied even to some participants who were found not to be carriers for an ICC after PGT because of worry and guilt for their family members who were found to be carriers (Whyte et al., 2016) or remain untested (MacLeod et al., 2014). Loneliness (Andersen et al., 2008, Bratt et al., 2012, Bratt et al., 2013, Geelen et al., 2011, Geelen et al., 2012, Meulenkamp et al., 2008, Smets et al., 2008), injustice and loss of freedom choosing leisure time activities, education and future professions are mentioned in some of the studies as having the most impact, particularly in younger participants (Bratt et al., 2012, Etchegary et al., 2016, Meulenkamp et al., 2008).

Those who have had to give up sport, where it was an important social context for them, as well as a personal interest or hobby, were affected the most. Grief resulted from this loss and was a dominant emotion in the first stages of receiving their results (Bratt et al., 2012). Some participants just refused to accept the lifestyle recommendations and carried on with their sport (Etchegary et al., 2016, Hendriks et al., 2005a) but others avoided certain activities because it worried their partners (Andersen et al., 2008). This consideration for how a participant's behaviour towards their status affected other family members extended to young ICC carriers who also expressed concern for their parents who become very agitated if they forgot their medications (Meulenkamp et al., 2008).

Sometimes, screening and/or PGT can bring about unexpected results, and this also caused emotional distress. In two studies, it was clear that patients who had an initial normal cardiac screen struggled when they were found to be a genetic carrier for an ICC (Ormondroyd et al., 2014, Smart, 2010). One participant described her feelings as 'really and truly shocking, I was absolutely horrified…' (Ormondroyd et al., 2014). Renewed mourning occurred in a family when genetic testing clarified that their deceased father

was the likely carrier of LQTS when it was always thought that their mother passed away due to this condition (Hendriks et al., 2005a, Hendriks et al., 2008).

In contrast, there were participants who felt that being a carrier was like having any other trait such as having red hair and therefore reported that their results had minimal emotional impact (Whyte et al., 2016).

Impact of symptoms, devices, and medications

Participants also expressed the need for more support regarding the management of and living with a diagnosis of an ICC. The internal cardioverter defibrillator (ICD), which is used for those with ICCs at high risk for dangerous arrhythmias, was a specific source of anxiety because of the practical implications of not being able to drive for a period of time and embarrassment of having visible signs of an implant (Andersen et al., 2008). Some worried about the ICD functioning properly (Andersen et al., 2008) whilst others were confident that devices would optimise their cardiac function (Meulenkamp et al., 2008) and were grateful it was implanted (Etchegary et al., 2016). Participants felt that they could have done with more time and information before the ICD was implanted (Andersen et al., 2008).

Patients were also confused about their symptoms in terms of recognising what was typical of the disease itself and what could be side effects of medications (Andersen et al., 2008); and were not clear about the consequences of not taking their medications or adhering to lifestyle changes (Bratt et al., 2013, Meulenkamp et al., 2008). Depression and fatigue were mentioned as a side effect of medications (Bratt et al., 2012).

Fear of death and dying due to the ICC was apparent (Bratt et al., 2012, Smart, 2010). However, others explicitly stated that this did not worry them because it is a normal part of life and death due to an ICC was perceived as relatively painless based on what they knew of their relatives who died suddenly (Andersen et al., 2008). Some participants

were also reassured because they were asymptomatic and believed that they were on the lower end of the spectrum in terms of the severity of the disease (Etchegary et al., 2016, Ormondroyd et al., 2014).

The ripple effect on the family

Following a diagnosis or an ICC carrier result, the next step in terms of family care is informing first degree relatives of their risk. Direct contact with relatives about this information was highly favoured although participants were aware that this might not be straightforward in some family circumstances (Ormondroyd et al., 2014, Whyte et al., 2016). Contact via a third party (Ormondroyd et al., 2014) as well as the use of social media (Whyte et al., 2016) was also considered as most felt that they would rather know about a possible risk than not and in some cases this facilitated a network for communication with distant family members. Some participants felt informing other relatives required further consideration. Those who seemed ambivalent about their own testing or did not have close ties with the family also expressed ambivalence about how relatives would react if they were informed of their risk (Smart, 2010, Whyte et al., 2016).

Some participants stated that some at-risk relatives did not have to be informed. Some carriers expressed that relatives who do not have or do not plan to have children may not even need to be informed that they can avail of PGT (Whyte et al., 2016). Elderly relatives (in the case of older adults and their parents) may be shielded from their risk information as screening would mean that they would worry unnecessarily about the participant's health and feel guilty about passing on an ICC (Ormondroyd et al., 2014). Furthermore, as ICCs occur in the young, it was rationalised that older relatives would have to start worrying about their own health when it is likely that by virtue of their longevity, they will continue to remain as they are even if they were found to be a carrier for an ICC (Smart, 2010).

3.5.3.3 Analytical theme 3: Strategies to cope with the results of the screening/genetic testing

Individual coping

Most studies discussed a transition and adjustment to living with the results of screening/genetic testing. After the initial period of shock and distress, which lasted from weeks to months, participants found ways to cope with their results. Strategies included a problem-focused approach to ensure participants had the information (Geelen et al., 2012), including online information (Whyte et al., 2016), to take necessary precautions alongside an emotion-based strategy to avoid bitterness about matters that they cannot change (Andersen et al., 2008, Meulenkamp et al., 2008), and having faith in medicine (MacLeod et al., 2014). A key task was adjusting to the lifestyle changes recommended and once this was mastered, there was a reorientation to new reference frames in life (Bratt et al., 2012) and gaining a sense of control (Ormondroyd et al., 2014). This enabled them to strive to reduce complications, see their situation in a positive light to enable them to move forward with hope and faith (Bratt et al., 2012).

Family and peer support

Support from their own families was deemed essential to help patients cope with their situation (Bratt et al., 2012, Manuel and Brunger, 2014, van der Werf et al., 2014, Whyte et al., 2016) but it is interesting to note that in Whyte et al.'s (2016) study, females were comfortable with sharing their concerns with relatives of both sexes whilst males would prefer to take an indirect approach or not share the information at all with other male relatives. Furthermore, communicating their health status beyond the immediate close family or friendship group was not common (MacLeod et al., 2014). Peer support from other patients/families with the same condition or experience was mentioned as something that could counteract the isolation and loneliness felt in rare conditions such as ICCs (Andersen et al., 2008).

Clinician support

Medical consultations became a source of security and reassurance as participants felt that any complications can be recognised and treated promptly (Bratt et al., 2012). Patients understood that the clinical team may not have all the answers and there may be differences amongst the clinicians, but the consultations were viewed as an act of support (van der Werf et al., 2014).

3.5.4 Synthesis 3: Quantitative and Qualitative Studies

The overall synthesis was guided by the aspects of care embedded within the review aims and objectives; and facilitated by a matrix with the juxtaposition of the findings from the parallel synthesis (Table 4). This facilitated a constant comparative analysis of the narrative findings from the quantitative studies and the thematic analysis from the qualitative studies. Concurring and conflicting evidence were identified, alongside areas of care that needed improvement.

Table 4 Synthesis of findings from quantitative and qualitative studies and recommendations

Key aspects of care for at-risk family members undergoing cardiac screening and/or PGT	Evidence of at -risk family members' experiences and views as they undergo cardiac screening and/or predictive genetic testing (PGT)		Reviewer recommendations
	From quantitative studies	From qualitative studies	
Clinic structure	Specialist inherited cardiac conditions (ICC) clinics were highly favoured for expertise and there was high satisfaction for the patient-clinician relationship and the experience of genetic counselling. A low proportion of patients attended follow-up	Specialist ICC clinics were viewed favourably for expertise for timely consultations. Genetic counsellors were highly regarded but more tailoring for young people was recommended	Consistent with established ICC guidelines specialist ICC clinics should be the gold standard for delivering care for ICC patients and their families. Further research is needed on the interaction and impact of other clinicians within the ICC multi-disciplinary team (MDT) aside from the genetic counsellor and medical staff.
Diagnostic or carrier status for an ICC	Lower quality of life (QoL) and depression is associated with a diagnosis of an ICC. There was conflicting evidence for emotional distress, nevertheless, high levels of anxiety and distress were experienced when screening and/or PGT results were received.	Uncertainty during the waiting period for results caused increased anxiety. The insight a test result gave to one's health was helpful and positive no matter what it was. Nevertheless, grief, high anxiety, shock, distress was experienced, especially when an ICC diagnosis or carrier status was confirmed.	Assessment of psychosocial needs should be done in a timely manner. Uncertainty impacts psychological status almost as much as a diagnosis or being a carrier for an ICC. The timepoint at which results are received is critical for providing psychosocial support and those who are diagnosed or found to be a carrier for an ICC; or received uncertain screening results, require additional input.

Key aspects of care for at-risk family members undergoing cardiac screening and/or PGT	Evidence of at -risk family members' experiences and views as they undergo cardiac screening and/or predictive genetic testing (PGT)		Reviewer recommendations
	From quantitative studies	From qualitative studies	
	Ambiguous screening results are associated with increased anxiety.		
Signs and symptoms for an ICC	There were higher incidents of depression and higher levels of anxiety in those with arrhythmic events, and signs and symptoms of an ICC. The presence of symptoms; and the uncertainty and the possibility of sudden cardiac death impacts on QoL and psychological distress.	The absence of symptoms was reassuring but when present, there was confusion as to the cause, whether this was due to the ICC or not. Consistent with the quantitative studies, fear of death and dying was prevalent.	Adequate health education should be provided about signs and symptoms of ICCs and a plan of action should they occur. An exploration of the risk assessment for sudden cardiac death and patient perceptions should be undertaken.
Medications and devices	Not mentioned in the studies.	Although, the internal cardioverter defibrillator (ICD) was thought of as protective, there were many concerns surrounding the ICD from pre-implantation to rehabilitation and the profound psychosocial impact this has.	Comprehensive preparation prior to device implantation and aftercare are cardinal aspects of ICD therapy. Adequate health education should be provided regarding medications used in ICCs.

Key aspects of care for at-risk family members undergoing cardiac screening and/or PGT	Evidence of at -risk family members' experiences and views as they undergo cardiac screening and/or predictive genetic testing (PGT)		Reviewer recommendations
	From quantitative studies	From qualitative studies	
		The consequences of not taking prescribed medications were unclear and they were felt to have unpleasant side effects and even cause depression.	
Lifestyle changes	Physical QoL and avoidance of activities are impacted by physical co-morbidities and the perception of poor health & limitations in daily activities.	A sense of injustice and loss was felt when lifestyle changes were imposed, particularly in sporty individuals, however, the consequences of non-adherence seemed unclear. Financial and insurance implications of a confirmed ICC diagnosis or carrier status contributed to emotional distress.	A thorough exploration and tailoring of advice for alternative activities considering the patients' risk assessments and co-morbidities, alongside interests and social practices is recommended. Signposting to resources and/or engaging appropriate professionals for socio-economic support should be part of comprehensive care.

Key aspects of care for at-risk family members undergoing cardiac screening and/or PGT	Evidence of at -risk family members' experiences and views as they undergo cardiac screening and/or predictive genetic testing (PGT)		Reviewer recommendations
	From quantitative studies	From qualitative studies	
Family	Increased anxiety is associated with a close relationship with proband, other family members with anxiety, absence of partner; and worry about family members not taking up PGT.	The main motivation of screening or PGT is to benefit the family. There is high distress in those who experienced a tragic, sudden death in the family, have a close relationship to the deceased but familiarity with ICCs in the family normalised the process of screening. Family support was deemed essential to adjustment to the diagnosis and/or carrier status.	The family plays a central role in how patients approach screening and/or PGT; and how they are impacted by the process. Careful consideration of the family history, family dynamics; and garnering family support are essential to planning tailored care, with added support for those who have experienced sudden death in the family.
Autonomy	Perceived benefits of genetic testing to family outweigh possible personal negative consequences.	The perceived benefits for the family are the main motivation for cardiac screening and/or PGT, with only a minor consideration for personal consequences.	The family plays a central role in decision making in ICC healthcare. Working with patients to elicit their intrinsic motivations should be incorporated to support autonomous decision-making. Autonomy support should be incorporated in the process of

Key aspects of care for at-risk family members undergoing cardiac screening and/or PGT	Evidence of at -risk family members' experiences and views as they undergo cardiac screening and/or predictive genetic testing (PGT)		Reviewer recommendations
	From quantitative studies	From qualitative studies	
		Sentiments of having no choice and imposition of screening and/or PGT evident. Subsequent at-risk family members are expected to take up screening and/or PGT.	facilitating cascade screening and/or PGT
Adjusting to a long-term condition	The longitudinal studies demonstrated normalisation of anxiety and reduction of psychosomatic symptoms occurred over time.	Patients described individual coping strategies and other sources of support that helped them adjust to their diagnosis and/ or carrier status. Peer support was suggested to decrease feelings of isolation.	Patients apply coping strategies to help them master the key tasks for adjusting to their condition. Providing education, setting of goals, and exploring strategies with patients to achieve these tasks could enable a faster period of adjustment as well as harnessing family, peer, and other social group support. Patients should be signposted to patient support groups and/or interventions could be provided in a group setting to provide peer support.

The gaps identified in service provision were used to make the following recommendations which should be incorporated within a psychoeducational intervention to support at-risk family members undergoing cardiac screening and/or PGT:

- Patients prefer to be seen in a specialised multi-disciplinary ICC clinic as recommended by clinical guidelines. Genetic counselling provision is beneficial but requires tailoring, particularly for young people. The role of other members of the multi-disciplinary (MDT), such as the cardiac genetics nurse (CGN), who tend to be the first point of contact for patients in the UK setting; and how they might improve their practice, has not been elicited in the included studies.
- Psychosocial assessment must be done in a timely manner to address needs promptly. The period when results are given is a particularly critical time, not just for those diagnosed or identified as carriers for an ICC but also for those who have been given ambiguous screening results.
- Signs and symptoms and the prospect of sudden death from an ICC is a major source of anxiety and depression and therefore comprehensive health education giving a clear perspective on the risk assessment for sudden death and a plan of action for signs and symptoms must be incorporated in the package of care.
- Education and psychosocial support should be provided around medication and device management. The ICD is a particular concern and patients who are candidates for implantation should be enrolled in the comprehensive pathway recommended (Dunbar et al., 2012) which should run in parallel to ICC services.
- The restriction of physical activity because of an ICC diagnosis or carrier status has a profound effect particularly for those where it is a major part of their social life. This may be imposed by clinicians or self-imposed due to perceived limitations or poor physical health. Whilst general guidelines exist according to type of ICC on exercise and activity restrictions, tailoring of advice is needed as patients differ in their risk assessments, preferences and may have co-morbidities.

- All aspects of the family - history, dynamics, sources of support, subsequent at-risk members -should be incorporated when assessing and planning patient care. A history of sudden death should also be an indicator that the patient is likely to need extra support.
- Decision-making in ICCs is mainly based on family reasons and whilst most do not regret their decisions, support and opportunities should be provided to help patients make autonomous choices. This support for autonomy should extend to the process of facilitating cascade screening or PGT for subsequent relatives.
- As time passes, patients adjust to their ICC diagnosis and/or carrier status. This period may be hastened if coping strategy support is provided and access to support groups is facilitated in a timely manner.

3.6 Discussion

The 22 studies included in this review were heterogeneous in design and setting, nevertheless, the parallel and combined synthesis of the quantitative and qualitative studies provided a comprehensive summary on the research so far on the experiences and psychosocial impact of cardiac screening and/or PGT for family members at-risk for ICCs. The review also sheds light on how patients cope when they are diagnosed and/or found to be a genetic carrier for an ICC.

This review identified that the main motivations for individuals to pursue cardiac screening and/or PGT is to ensure the maintenance of their own health and their kin. This is consistent with the findings in PGT in other genetic health conditions (Claes et al., 2004, Clark et al., 2000, Lammens et al., 2010, Lerman et al., 2002) and also in ICC probands seeking genetic testing (Andersen et al., 2008, Erskine et al., 2014, Etchegary et al., 2015, Geelen et al., 2012, Geelen et al., 2011, Smart, 2010). Most participants engaged freely but pressure from parents and spouses; and the influence of health care providers were reported and in some, this caused resentment. This is a similar finding in

a review of presymptomatic testing of young adults (Godino et al., 2016) and the challenge for health professionals would be to ensure that autonomous decision making is facilitated in the context of multiple issues. This includes the health professionals' own awareness that taking up screening or testing could prevent adverse outcomes for the individual and their family (Bartels et al., 1997).

In participants who decided not to pursue screening/testing, mostly in the setting of having had some contact with a health professional, it was worrying that there was a perception that this will not facilitate preventative measures (particularly for HCM) but indeed it can (Elliott et al., 2014). Furthermore, genetic discrimination was mentioned as a reason for not pursuing genetic testing in Geelen et al.'s (2011 & 2012) studies in a context where there are many laws to prevent this therefore, it is likely that these fears and misunderstandings may have stemmed from other family members' early experience of no perceived benefit of screening and/or PGT and discrimination. Geelen et al. (2012) recommended that counselling must not be limited to reassurance that legislation exists but also to explore the family's perceptions of genetic discrimination and coping strategies that could be a barrier to genetic testing.

Based on the quantitative studies, this review has demonstrated that overall, that the process of cardiac screening and/or PGT does not confer a great risk for a lasting adverse psychological impact. This is consistent with other reviews on genetic conditions (Broadstock et al., 2000, Godino et al., 2016, Heshka et al., 2008). However, patients generally experience increased anxiety at initial clinic attendance and when they receive their results; and along with the findings for depression and poorer QoL, this is not associated with carrier status per se. Communication in genetic consultations should focus on the psychosocial aspects of the inherited condition due to the predisposition to distress in certain families (Kissane and Parnes, 2014) and those described in this review (McGorrian et al., 2013), the specific emotional reactions described in the qualitative studies; and research showing that prior emotional status can be predictive of

subsequent psychological issues (Broadstock et al., 2000). However, it has been shown that consultations often focus on the physical and biomedical aspects of the genetic condition (Paul et al., 2015). A pre-test assessment of emotional state (Broadstock et al., 2000) and an initial exploration of possible reactions to initiating the testing process (Godino et al., 2016) may help tailor the conversation according to the needs of patients in order address ambivalence and psychosocial needs from the outset.

In living with the outcome of screening/testing results, a normal or abnormal outcome brings about different challenges. Whilst normal results bring relief, the 'survivor guilt', doubt and anger described is a common finding in genetic conditions, as they now must adjust to a new situation and give up their identity of an at-risk person (Graceffa et al., 2009, Metcalfe et al., 2008, van't Spijker and ten Kroode, 1997).

For those diagnosed with an ICC, the severity of signs and symptoms and accompanying medical interventions, particularly the ICD; lifestyle advice; and the perception of risk for sudden death caused a negative impact on QoL and increased anxiety. However, as in other genetic conditions, a transition to the mastery of the adjustments required and development of coping mechanisms were shown to facilitate as sense of control and decrease in psychological distress (Berkenstadt et al., 1999, Biesecker and Erby, 2008, Hoedemaekers et al., 2007). Individuals are naturally orientated toward growth and well-being and will interact with the environment to achieve these aims (Deci and Ryan, 2000). Therefore, there is a huge potential to optimise both the social and healthcare environment for fostering this motivation (Biesecker and Erby, 2008, Davey et al., 2005, La Guardia, 2017).

Whilst participants strive to maintain their own health, the family looms large in terms of the next steps following an ICC diagnosis or confirmation of carrier status. There is a strong desire to inform and protect family members who may be at risk, and this extends to planning for future offspring. This sense of 'genetic responsibility' prevails from the

time participants were making their testing/screening decisions and is a common theme in studies that deal with communicating genetic risk to family members and reproductive decision-making (Leefmann et al., 2017). This issue is now becoming more acute in ICCs due to greater access to screening and genetic testing, and availability of effective disease management and prevention of sudden death. There is a risk that this overriding desire to make sure relatives have taken up screening/genetic testing by both patients themselves or genetic practitioners could impinge on autonomy (Huibers and van`t Spijker, 1998). In contrast, there were views from participants that there were family members that they would not consider as needing to know about their genetic risk, such as older relatives. This does not go against the principles of genetic responsibility as its premise is on the idea of 'do no harm' (Leefmann et al., 2017), however, even older individuals can benefit from therapies if required and in autosomal dominant conditions such as ICCs, they are usually key in unlocking the opportunity for screening/testing in extended family members (Elliott et al., 2014).

The findings from this review show that risk communication may not be straightforward and requires going beyond the simple giving of information to encompass the appreciation of patients' value systems. Indeed, counselling-orientated interventions focused on this process are specifically designed to promote a supportive environment for decision-making rather than a prescriptive approach (Eisler et al., 2016, Hodgson et al., 2016).

This review illustrates how healthcare services have evolved following the increased knowledge and advances in the field of ICCs over the years. From reports of very poor experiences with health practitioners from the early 1980s to greater satisfaction with the current specialist, multi-disciplinary clinic models which is the recommended model of care today (Elliott et al., 2014, Fellmann et al., 2019, Ingles et al., 2011, Priori et al., 2013). There was great satisfaction with the contribution and support from the genetic counsellors, however, tailoring of counselling to specific needs could be improved. In

young people, a comprehensive longitudinal counselling process (Waldboth et al., 2016) has been suggested which encompasses 'patient-to-offspring' risk communication and the young person's own decision-making on testing and risk management (Godino et al., 2016). Only the doctors and genetic counsellors were specifically mentioned in the studies as providing genetic healthcare, but multiple professionals are usually involved in the care of ICC patients, including specialist nurses, cardiac physiologists, and clinical psychologists (Arscott et al., 2016, Kirk et al., 2014, Watts et al., 2009). With hospital consultations identified as a source of support, this creates an opportunity to enhance access and continued follow-up; and to look at various facets of the service where care could be improved.

3.7 Implications for practice and further research

The major output of this review is a set of recommendations for improving the experience and clinical care of at-risk family members who are undergoing cardiac screening and/or PGT for an ICC based on the synthesis of evidence from existing qualitative and quantitative studies (Table 4). These recommendations serve as a springboard from which to conduct service improvement initiatives and further research into this population.

The experiences of family members at risk for ICCs as they go through cardiac screening and/or genetic testing has evolved since the 1980s. The advent of specialised, multi-disciplinary clinics and established clinical guidelines have facilitated comprehensive medical care and improved patient experience. This is now considered the gold standard of practice and any patient at risk for ICCs should be referred to these services. However, it is evident from this review that despite these improvements, there are gaps in service provision, particularly the need of more tailored health education and psychosocial support as patients undergo emotional upheaval in the initial stages of this process; as they adjust to their situation; and as they disseminate risk information to family members.

Furthermore, many patients decline screening/testing or get lost to follow up because of misconceptions and this may have consequences on the patients' and their families' long-term outcomes, including risk of sudden cardiac death.

In the current clinical setting, prioritising the patients' psychosocial concerns and values over the biomedical aspects of ICCs during clinician interactions, particularly those with the genetic counsellor and cardiac genetic nurse, may enable the team to tailor care accordingly and reduce anxiety through the timely implementation of interventions and referrals to appropriate services.

Future research in determining psychological distress prior to consultations may help in planning and anticipation of patients' healthcare needs during screening/testing and when results are received. Identifying the basic psychological needs of patients such as autonomy, competence and relatedness in the context of ICCs and whether these have been addressed could facilitate the development of interventions to fulfil these needs (La Guardia, 2017). Overall, the appreciation that the family unit and many other factors, such as perceptions about symptoms and risk of sudden cardiac death; influence the outcomes of this patient group and should be incorporated in any future study.

3.8 Limitations

The synthesis of the wealth of findings from both quantitative and qualitative studies in this review enabled a comprehensive view on the patient experience and more robust recommendations however, it is also important to acknowledge possible issues with the methodology. Conducting mixed methods systematic reviews is an evolving method and whilst every effort has been made sure to ensure fidelity to the chosen systematic review method (adherence to coding and analysis standards, utilisation of a matrix), it was modified to accommodate non-experimental quantitative findings. This, alongside the

diversity of the studies, precluded that the review would not have any pooled data from the meta-analysis of RCT findings found in conventional reviews.

It is not surprising that HCM and LQTS have a greater representation in this review as HCM is the most common of the ICCs and both have much more established guidelines for screening and genetic testing than the other ICCs. Yet, both conditions are still relatively uncommon in clinical practice, hence, the combination of several ICCs in the studies included. It is possible that as more studies are done on each disease entity, more specific recommendations can be made than those presented in this review. Furthermore, participants in the studies tended to be a mixture of probands, at-risk relatives and significant others (spouses or parents) of various ages and if more focused studies are conducted on greater numbers, these may yield different findings.

Another possible limitation for this review was that only studies published in English and in peer-reviewed scientific journals were included. Therefore, this may not include the data available in the grey literature and unpublished manuscripts. The studies included were all conducted in developed countries, mainly in Western Europe, the US and Australia; and may not reflect the experiences of those with a different socioeconomic and cultural background. All the studies which included participants who decided not to pursue screening/PGT were likely to have had some contact with a healthcare professional therefore, there is hardly any representation from at-risk relatives who chose not to engage with the healthcare system.

3.9 Implications of the review to the development of the psychoeducational intervention

This review established the preliminary evidence base for developing the psychoeducational intervention. By eliciting the experiences and gaps in service

provision for at-risk patients undergoing screening and/or PGT for ICCs, recommendations for improving this can be incorporated in any proposed intervention. The components of the psychoeducational intervention are emerging in the form of the contents of the psychological component which should incorporate early assessment of psychological status and needs. The exploration of perceptions, values and preferences, autonomy support; and harnessing support from family or social groups are also key ingredients. For the health education component, information about the management of signs and symptoms, indications for medications/devices and dealing with side effects should be incorporated.

A wide range of outcome measures were used in the included studies, mainly focused on psychosocial status and patient satisfaction and these could be used in measuring the effectiveness and acceptability of an intervention. However, outcome measures that have specificity to the intervention and relevant to at-risk family members must be considered further.

The core themes generated from this review: family, psychosocial adjustment, and autonomy, contributed to the development of the theoretical framework for the psychoeducational intervention which is discussed in detail in Chapter four. Whilst tailoring health advice and counselling was emphasised, the form and manner by which psychoeducational interventions should be delivered was not elicited in this review and therefore a further study was conducted to establish this as reported in Chapters 5-7.

Chapter 4: Identifying and developing theory

4.1 Introduction

According to the MRC Complex Intervention Framework, developing a theory-based psychoeducational intervention will have the advantages of eliciting the mechanisms of the intervention, identifying the targets and the techniques to deliver the intervention; and develop outcome measures (Craig et al., 2008). However, the MRC Framework does not illustrate the practical steps to enable researchers to select the appropriate theory and it tends to be left to the expertise of the researchers, the research questions raised and the context of the specialist field (Hawe, 2015). This chapter describes how Self-Determination Theory (SDT) was chosen as the theoretical framework of the psychoeducational intervention by critically appraising the theoretical underpinnings of psychoeducational interventions in cardiovascular and genetic healthcare; and the emerging ICC interventions (Lippke and Ziegelmann, 2008), alongside the findings of the preceding systematic review (Chapter 3).

4.2 Theoretical perspectives of psychoeducational interventions in cardiovascular and genetic healthcare; and inherited cardiac conditions

4.2.1 Psychoeducational interventions in cardiovascular healthcare

Health is a pre-requisite to attaining other goals in life. Yet the quest for health is often derailed by habits and lifestyles (Ryan and Deci, 2017). Behavioural interventions in healthcare are typically focused on adopting healthy habits for disease prevention and adherence to therapeutic recommendations (Kwasnicka et al., 2016). In the earliest reported systematic review of psychoeducational interventions in cardiovascular healthcare, it was concluded that the lack of references to a theoretical model in the included studies limited the ability to shed light on the mechanisms and effective components of the interventions (Dusseldorp et al., 1999).

In the more recent systematic reviews on this topic, it was noted that establishing a theoretical basis for the interventions targeting behavioural determinants to improve the outcomes for improving chest pain, smoking cessation, exercise uptake, weight loss and a healthy diet; was becoming the standard (Aldcroft et al., 2011, Huttunen-Lenz et al., 2010, McGillion et al., 2008). The most common behavioural change theories used in the primary studies in these three systematic reviews, where specified, were the Transtheoretical model of behaviour change (Prochaska and DiClemente, 1983) or the application of stages of change (Bolman et al., 2002, Izawa et al., 2005, Ockene et al., 1992, Reid et al., 2003), Self-efficacy theory (Dornelas et al., 2000, Feeney et al., 2001, Johnson et al., 1999, McGillion et al., 2014, Taylor et al., 1990, Yates et al., 2005), Interactionist role theory (Dracup et al., 1984), Cognitive behavioural theory (Lewin et al., 1995, Lewin et al., 2002) and Marlatt-Gordon's Cognitive-Behaviour Model (Smith and Burgess, 2009). These interventions were specifically aimed at encouraging lifestyle changes and decision-making in individuals with overt heart disease, hence, the use of theoretical frameworks that provide knowledge, skills and encourage action for stopping or changing a harmful health behaviour such as smoking and a sedentary lifestyle.

4.2.2 Psychoeducational interventions in genetic healthcare

In psychoeducational interventions in genetic healthcare, the theoretical frameworks used included: Theories of stress and coping (Halbert et al., 2004, Katapodi et al., 2018), Cognitive-social theory (McKinnon et al., 2007, Roussi et al., 2009); Health Belief Model (Kasting et al., 2019) and Mischel's (1988) Illness uncertainty theory (Maheu et al., 2015). These interventions were mainly aimed at women affected or at high risk for hereditary breast cancer and were focused on improving knowledge and skills for accessing clinical services, decision-making with respect to genetic testing, providing support following the result, allaying stress, and anxiety; and facilitating family communication. In contrast to the psychoeducational interventions in cardiovascular healthcare, rather than focusing on stopping or changing a harmful health behaviour, the

participants were encouraged to adopt health protective practices such as self-breast examination and relaying risk information to family members. Thus, the theories used were rooted in identifying stressors and coping mechanisms alongside health education. Despite this distinction, Prochaska & DiClemente's (1983) Transtheoretical model of behaviour change was also used in one study which was focused on a behaviour change to increase the uptake of genetic counselling amongst breast cancer survivors (Kasting et al., 2019).

An intervention grounded in Family Systems Theory has been used to support family communication about the genetic condition, and to assist family adaptation to living with the condition or the risk of it developing. The Multi-family discussion group (MFDG) psychoeducational intervention developed by Eisler et al. (2017) for a range of genetic conditions, is based on the concepts from systemic therapy, cognitive behaviour therapy and group therapy (Asen and Scholz, 2010). The outcomes for this intervention were in keeping with those already mentioned in genetic healthcare but with a focus on harnessing the strengths of the family unit to promote coping and adaptation.

4.2.3 Psychoeducational interventions in inherited cardiac conditions

In the psychoeducational interventions developed for the ICC population so far, Hodgson et al.'s (2016) study specified a theoretical framework which was the Reciprocal Engagement Model of Genetic Counselling (REMGC) (Veach et al., 2007) which postulated that the interaction between the proband for an ICC and the genetic counsellor impacts on the uptake of genetic services. Kinnamon et al. (2107) cited Self-Regulation theory (Cameron and Leventhal, 2003) as the theoretical framework for the development of the Family Heart Talk intervention as modelled on a previously successful family communication aid developed for preventative behaviours melanoma survivors (Bowen et al., 2017).

4.3 Applying the theoretical frameworks in the context of family members at-risk for inherited cardiac conditions

As no psychoeducational intervention has been specifically developed for family members at-risk for ICCs to date, the theoretical frameworks mentioned in Section 4.2 should be considered to inform the psychoeducational intervention being developed in this research. However, it is important to critically analyse these in the context of this research to determine whether they would be appropriate (Lippke and Ziegelmann, 2008).

4.3.1 Knowledge and skills for effective coping

Whilst some theoretical frameworks mentioned thus far may differ in names and their proponents, they are inter-related as some are based on another or an amalgamation of several frameworks. The Self-efficacy theory appears to be a major construct of a number of theoretical frameworks as it has its roots in the belief that deviant behaviour is not a disease in itself but rather an interplay of personal, behavioural and environmental factors; and interventions can be delivered by various practitioners and in different settings (Bandura, 2004b). Within this theory, self-efficacy is the belief in one's own ability to be successful in achieving something and individuals will tend to act if they believe they can accomplish the task. Conversely, if individuals think they will fail, it is unlikely that they will act. It is, therefore, important to improve self-efficacy by providing knowledge and experiences for the mastery of a task, role models for vicarious experiences, verbal persuasion; and support in reducing stress, anxiety and worry (Bandura, 1997).

In the Transtheoretical model of behaviour change (Prochaska and DiClemente, 1983), self-efficacy is a major consideration as it heralds the six stages within which behavioural change occurs. These stages are defined by an individual's readiness to take action and interventions are tailored according to the stage the person is in (Marcus and Simkin,

1994). For example, in the early stage of ‘precontemplation’, there are low levels of self-efficacy or confidence in one’s belief to make the desired changes and the disadvantages of behaviour change are thought to outweigh the advantages. The intervention strategy for this stage would therefore focus on a process of change that involves ‘consciousness raising’ to increase awareness and provide information on how to enable this change. Whereas in the later stages of change such as ‘action’ or ‘maintenance’, the individual has a much higher level of self-efficacy to change their behaviour and resist temptation, therefore, seeking ‘helping relationships’ would be the appropriate process of change in terms of intervention.

4.3.2 The personal perspective

The Self-efficacy theory is also a major construct of Cognitive social theory (Bandura, 2004a) which is in turn, the basis of Marlatt and Gordon’s Cognitive behavioural theory (1999). The work of Marlatt and Gordon is specifically tailored to relapse prevention in alcohol addiction and identifies the high-risk situations that precipitate a relapse that threaten self-efficacy. This includes emotional states (negative and positive) and social situations (Larimer et al., 1999). In these latter two theories, there is an emphasis on how an individual perceives and interprets a situation and the degree of self-efficacy to carry out the necessary behaviour change to improve this situation if required. Cognitive behavioural theory also posits that the perception of events is the major influence in people’s behaviours and emotions (Beck and Beck, 1995). In clinical practice, the ‘cognitive model’ is used to understand the causes, precipitants and influences of a problem to make sense of the person’s experience and facilitate a mutual understanding of their difficulties (Fenn and Byrne, 2013).

Furthermore, the Stress and coping theory is also consistent in utilising a personal perspective as the individual conducts an appraisal of the environmental stressors. Coping strategies are then deployed as the cognitive behavioural effort to manage the demands of the situation (Folkman et al., 1986). Uncertainty is often cited as a cause of

stress in genetic healthcare; however, the experience of uncertainty is initially neutral until it is appraised. This is where the personal perspective is emphasised once again in the Health Belief Model (Becker, 1974) and the Illness uncertainty theory where uncertainty is assessed as a danger or opportunity; and coping mechanisms are deployed accordingly (Mishel, 1988). In these theories, it is noted that the coping strategies required for adaptation may exceed the resources of the individual.

In the work of Miller et al. (1996) applied to the work of Roussi et al. (2009), they extended Self-efficacy theory and the Cognitive social theory by concluding that the perceptions/cognitions and affects triggered when processing health information have relatively stable structures and that this can guide which of the two cognitive-affective styles they proposed will be used when responding to health threatening information. These are the 'high-monitoring' cognitive-affective style where there is deeper attention and scanning for threatening signs and therefore should result in better coping; and the 'low-monitoring' cognitive-affective where there is often distraction and threatening signs are ignored which leads to negative emotional responses and poor coping (Miller et al., 1996).

4.3.3 Social relationships and interactions

Whilst the other theoretical frameworks touched on the importance of the relationships and the interactions of the patient, their significant others, and the clinician, this is given much more focus in the Reciprocal Engagement Model of Genetic Counselling (REMGC), Interactionist role theory; and the concepts brought into delivering MFDG Interventions. With a central tenet of the relationship between the genetic counsellor and the patient as integral to the counselling process, the REMGC gives more emphasis on how an effective clinician-patient relationship fosters health education and optimises clinician and patient attributes. This is said to bring about the outcomes of genetic counselling facilitating autonomous patient decision-making and adaptation (Veach et al., 2007).

The Interactionist role theory states that family, friends and clinicians are important in helping patients adopt a 'compliance role' within their self-concept by acting as complementary/supporting roles; giving cues and serving as a point of reference for evaluation of compliance to a therapeutic regime (Dracup and Meleis, 1982). The MFDG interventions take the role of the family even further by using the Systems theory perspective which views the family as an ecosystem with different parts working together towards a coherent whole. By bringing in the concepts from group therapy and cognitive behavioural therapy, the family groups are facilitated to support and learn from each other on the strength of their common experiences with genetic conditions (Asen and Scholz, 2010). This is a promising model for delivering a psychoeducational intervention in the group at-risk for ICCs, however, the challenge remains in adopting this into routine healthcare (Eisler et al., 2017).

4.3.4 Applicability to the current project

It is evident from the studies for psychoeducational interventions in cardiovascular and genetic healthcare that these interventions have a common ground in utilising theoretical frameworks which facilitate increasing knowledge and skills in coping with the health condition. However, in cardiovascular diseases, interventions are focused on improving lifestyle choices to minimise risk factors that could worsen the condition whilst in genetic conditions, these are mainly deployed to support decision-making to take up health protective practices, decrease stress and anxiety and facilitate family communication. Based on the findings in the systematic review in Chapter 3, all these features are required in developing an intervention for the population at-risk for ICCs.

Health education and coping skills; the role of the family, other social groups and the clinical team have all been identified as essential aspects of care, as well as support for both stopping or changing an 'unhealthy' behaviour (some forms of exercise are discouraged in certain ICCs) and taking up health protective behaviours such as attending yearly monitoring. The Cognitive social theory appears to be the most

comprehensive in these aspects, alongside the prioritisation of patient-centred care based on the emphasis on the individual perspective. However, addressing autonomy support needs was not implicit as required in the recommendations of the systematic review. Autonomy is a tenet in the Reciprocal engagement model of genetic counselling, but this was limited to decision-making within a genetic counselling session.

In the context of this research, the Transtheoretical model of behaviour change and Marlatt and Gordon's Cognitive behavioural theory appear to be the least applicable as their evidence base was established in studies on addictions which is strikingly different from the nature of an inherited condition. Furthermore, in the Transtheoretical model of behaviour change, social support is thought to be more appropriate in the later stages of change and the systematic review has indicated that this may need to be drawn on at any stage.

This critical analysis of theoretical frameworks used in the psychoeducational interventions in cardiovascular and genetic healthcare has not identified a unifying theoretical framework for developing a psychoeducational intervention for patients at-risk for ICCs. This has prompted exploring the wider field of behavioural psychology to find a theoretical framework that captures not only the need for improving self-efficacy within a social context but also to facilitate autonomy support.

4.4 Proposing Self-determination theory as the theoretical framework for the psychoeducational intervention for family members at risk for inherited cardiac conditions

4.4.1 Self-determination theory

Self Determination Theory (SDT) is a theory of motivation integrating the two viewpoints that individuals have natural, constructive tendencies and that there is a need for supportive social-contextual factors to develop a coherent sense of self. SDT proposes that the three innate human psychological needs of autonomy, competence and relatedness must be satisfied within the social context to promote intrinsic motivation (Deci and Ryan, 2002).

Autonomy is the degree to which individuals feel volitional and take ownership of their behaviour. Competence is the extent to which they feel they can reach their goals and expectations. Relatedness is the extent to which they feel they connect to others in a warm, positive and interpersonal manner (Ryan and Connell, 1989). The mention of a social context recognises that significant others/family members also play a key role in SDT. An environment which supports the basic psychological needs will promote engagement, mastery and synthesis; however, when they are thwarted, this will be detrimental to motivation, growth, integrity, and well-being.

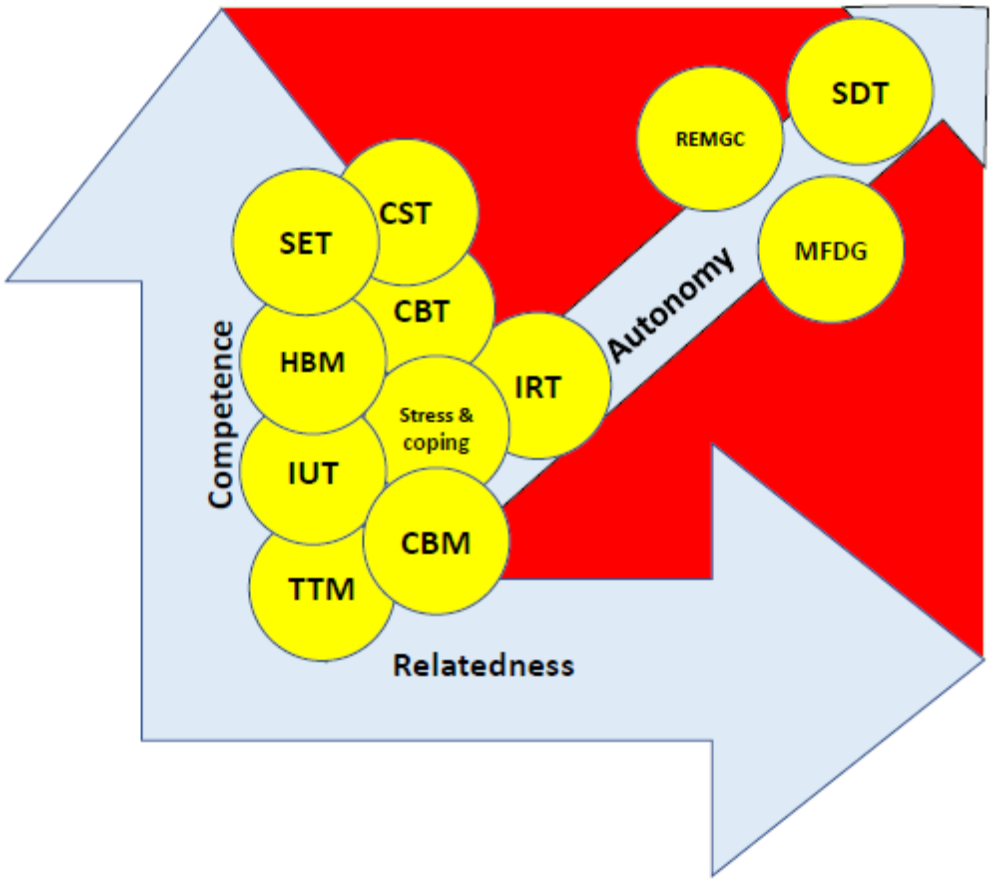
Self-Determination theory suggests that the degree of motivation exists in a continuum according to how the three psychological needs are met. Intrinsic motivation is the highest form of motivation on the self-determination continuum where there is ownership of a decision or behaviour, and the individual does not feel controlled or pressured; engagement is brought about by enjoyment and interest. In contrast, external motivation is acting when prompted by others, for gaining a reward or avoiding punishment. At the extreme end of this spectrum is amotivation where an individual has no intention to act or acts passively (Williams et al., 2004). The more intrinsically motivated an individual is,

the more likely it is that lasting behaviour change will occur, and a greater sense of well-being is fostered.

4.4.2 The rationale for Self-determination theory in the development of a psychoeducational intervention for family members at risk for inherited cardiac conditions

The basic psychological needs proposed by SDT addresses all the aspects of care that have been identified in the systematic review in Chapter 3 that need to be incorporated for a psychoeducational intervention for patients at-risk for ICCs. Furthermore, it provides a comprehensive theoretical framework by combining the aspects of those that have been used in cardiovascular and genetic healthcare that are applicable to this population. Figure 8 summarises the theoretical frameworks discussed thus far in the context of the basic psychological needs. Improving self-efficacy is built into the concept of 'Competence' as this encompasses the health information and coping skills that individuals need to have confidence in managing their health. The importance of the family-in both experiences and relationships; the interactions with health professionals and other social connections are captured in 'Relatedness'. The need to support intrinsic motivation in decision making across all aspects of ICCs is emphasised in 'Autonomy'.

Figure 8 Summary of theoretical frameworks



Abbreviations: Cognitive Behavioural Model-CBM, Cognitive Behavioural Theory-CBT, Cognitive Social Theory-CST, Illness Uncertainty Theory-IUT, Health Belief Model-HBM, Interactionist Role Theory-IRT, Multi-Family Discussion Group-MFDG, Reciprocal Engagement Model of Genetic Counselling-REMGC, Self-Efficacy Theory-SET, Self-Determination Theory-SDT, Transtheoretical Model of behaviour change-TTM

Alongside functioning as a framework to identify components and targets for the psychoeducational intervention, SDT is also proposed to explain the phenomenon of adjustment and adaptation that occurs in at-risk ICC patients who are identified as genetic carriers and/or diagnosed with an ICC. This is due to SDT's basic assumption that satisfaction for competence, autonomy and relatedness is the essence of human thriving and contribute to indicators of wellness and vitality (Ryan and Deci, 2017).

Most SDT health interventions are focused on behaviour change in fields such as smoking cessation, physical activity, weight loss, diet, and adherence to medications (Gillison et al., 2019). Applying SDT to develop a psychoeducational intervention for

patients at-risk for an ICC is a novel approach and therefore the evidence base for SDT required greater scrutiny.

4.4.3 The evidence base for Self-determination theory

Self-determination theory has evolved over five decades through a top-down, empirical approach to test and validate SDT's theoretical propositions (Ryan and Deci, 2017). It is recognised as a key theory in underpinning maintenance of behaviour change (Kwasnicka et al., 2016) and the evidence for the efficacy of SDT-based interventions is growing with a number of RCTs in the health domain conducted internationally. Collectively, the efficacy of SDT-based techniques within health interventions were synthesised in a meta-analysis which showed that in 74 clinical trials, the SDT techniques used can impact on the theoretical mediators of health behaviour change. The effect sizes varied from a large effect size for autonomy support and satisfaction, moderate effect size for competence satisfaction to a small effect size for relatedness satisfaction and autonomous motivation. It was suggested that rather than using one particular technique, a combination of synergistic techniques is more favourable in creating a supportive environment (Gillison et al., 2019).

A more recent systematic review and meta-analyses of 56 articles describing 65 tests of SDT interventions found that the effect sizes were more modest at $d_+ = 0.23$ vs. 0.41 and this was felt to be impacted by publication bias and small sample bias (Sheeran et al., 2020). However, utilising the Meta-analytic structural equation model, the authors were able to elicit a more refined path analyses which showed SDT interventions promoted health behaviour change via increased autonomous motivation and perceived competence. To operationalise these SDT techniques in the ICC setting, several trials were studied in greater detail.

4.4.3.1 Exercise and lifestyle in healthy populations

Tailoring exercise and lifestyle advice are an important feature in the care of at-risk patients with ICCs. There are several related SDT-based intervention trials in this aspect that tested for efficacy and conducted evaluations of the SDT process model. To improve the uptake of physical activity a study incorporating an autonomy-supportive, well-structured and interpersonally involving teaching style in a 10-week exercise programme for university students showed that these SDT features led to a significant increase in relatedness and competence need satisfaction over time and better attendance rates compared to the control group whose perspective of the exercise class was not taken in (Edmunds et al., 2008). This is the first study involving a trial of an SDT-based intervention in a real-world setting providing evidence that such interventions are feasible and training of personnel to teach in an SDT-orientated style is possible.

A larger cluster RCT involving 13 centres with an exercise referral scheme showed that in a total of 347 participants, those who received an exercise referral intervention grounded in SDT, showed significant improvements in anxiety levels compared to controls at six months (Duda et al., 2014). Within the intervention group, physical fitness measurements also improved significantly during this period. Process modelling supported the hypothesis that the SDT intervention model provided autonomy support through the health advisor, need satisfaction and more autonomous motivation, enhanced engagement in physical activity and promoted wellbeing. Another RCT conducted in Canada for a physical activity counselling intervention showed that autonomy support and autonomous motivation was significantly higher in the intervention group at six weeks; and at 13 weeks, also showed higher levels of physical activity compared to the control group (Fortier et al., 2007). Consistent with the previous study, the path analysis for the SDT model, showed that autonomous motivation and perceived competence at six weeks predicted the physical activity for the experimental group.

A Portuguese RCT for a one-year SDT-based behaviour change intervention targeting overweight and mildly obese women (N=239) showed that those in the intervention group were highly successful in taking up moderate and vigorous physical activity and achieving weight loss at one year compared to controls (Silva et al., 2010). The intervention also showed significant effects on SDT-related variables such as higher self-regulation, exercise intrinsic motivation, perceived competence, and internal locus of causality.

In terms of medication adherence, an RCT for an intervention for smoking cessation designed specifically to support autonomy and competence using intensive counselling, regardless of intention to quit, was found to improve abstinence and adherence to medications to support smoking cessation compared to usual community care (Williams et al., 2006). This study also demonstrated that these two outcomes were facilitated through the internalisation of autonomous motivation and perceived competence through the analysis of the SDT process model.

4.4.3.2 Exercise and lifestyle in specific disease states

Whilst in the trials in relatively healthy individuals are a good indication that SDT-based interventions are effective in supporting a healthy lifestyle, there was also evidence that they are helpful in populations with conditions associated with physical limitations, which require much more input, such as in ICCs. In those with rheumatoid arthritis, an SDT-based intervention brought about a higher competence need satisfaction; positively predicting change in autonomous motivation which in turn positively predicted moderate physical activity at three months (Duda et al., 2015). In HIV-infected older adults with some physical limitations, participants who were randomised to an SDT-based physical activity counselling programme had a significant improvement in measures of physical function and levels of physical activity; measures of depression and QoL; and measures of autonomous regulation compared to the control group (Shah et al., 2016). Halvari et al. (2017) conducted an RCT for an SDT-based physical activity programme in Norway,

in 108 adults with both diabetes mellitus and coronary artery disease. The intervention group showed higher levels of performance of physical activity, blood sugar testing; higher levels of vitality perceived health; and decreases in HbA1c levels. This study also showed through path analysis that improved physical and mental health can be achieved using the psychological mechanisms of satisfaction of basic psychological/motivational needs, internalisation of autonomous self-regulation, and increased perceived competence (Halvari et al., 2017).

In contrast, an RCT of an eHealth intervention (SurvivorCHESS programmme) based on SDT to increase physical activity in colon cancer survivors with 144 patients in the intervention group resulted in increase in exercise over time for all participants but there was no significant difference for exercise uptake between the intervention and the control group (Mayer et al., 2018). Self-determination theory, including autonomous motivation and relatedness, was not associated with the outcomes, highlighting that more consideration would be required when applying SDT to purely app-based interventions.

So far, in genetic healthcare, the Engage-HD Physical Activity intervention, an SDT-based intervention with an individualised lifestyle approach to support physical activity in people with Huntington Disease (Busse et al., 2014) has undergone a feasibility RCT. Forty-six percent of eligible patients were randomised to either the physical intervention with an uptake of 82%, or the social intervention which had a 100% uptake. There were no evident treatment effects on physical function, however, the increase in self-efficacy for exercise and self-reported levels of physical activity for the physical intervention supported the predefined intervention logic model based on SDT (Busse et al., 2017).

4.4.3.3 Cardiorespiratory disease self-management

As in ICCs, serious medical conditions can affect family relationships therefore the concept of providing support to both patients and caregivers to improve outcomes are being explored in SDT intervention development. In advanced lung cancer a telephone

based SDT intervention delivered to patient and caregiver dyads proved feasible and showed significant improvements in anxiety, depression and caregiver burden compared to usual medical care delivered (Badr et al., 2015)

Two studies in North America used SDT-based interventions in an RCT for the cardiovascular disease population, specifically for those with heart failure, both involved family partners/caregivers and patient dyads in promoting self-care (Cossette et al., 2016, Dunbar et al., 2013, Stamp et al., 2016). Cosette et al. (2016) undertook a randomised pilot trial where post-discharge heart failure patients and their carers were offered a total of five encounters (the first was face-to-face with the dyad and the second with the caregiver only and follow up was done over the telephone) with a nurse trained in using SDT-based communication techniques individualised according to the dyad's needs. Despite issues with recruiting live-in caregivers for this trial and dropouts due to frailty in this population, the intervention was deemed acceptable and feasible; and had favourable outcomes for self-care in relation to heart failure, internal extrinsic motivation, intrinsic motivation, and caregiver's feeling in being able to give better support for the experimental group compared to the control group.

Dunbar et al. (2013) and Stamp et al. (2016) conducted a full RCT on the SDT-based intervention with these comparison groups: one receiving usual care using pamphlets; a patient family education group with heart failure patient and family partner dyads receiving a one-hour education session with a nurse and a group session facilitated by the nurse and a dietitian; and the family partnership intervention group with dyads receiving the latter sessions and two additional sessions focused on teaching the dyads on how to support each other's roles. Both the patient family education group and the family intervention group showed improved dietary sodium intake compared to the usual care group; there was no difference in terms of medication adherence amongst the groups (Dunbar et al., 2013). This study concluded that the family partnership intervention significantly improved confidence and motivation at four months whereas

there was no change in the other groups. Family functioning was also evaluated in this study using Family Assessment Device Questionnaire based on the McMaster Model of Family Functioning (Epstein et al., 1983), which conceptualises the organisation of families and their interactions. The results showed that those with better family functioning had more self-care confidence for diet and autonomous motivation for diet and medications (Stamp et al., 2016).

4.4.4 Quality appraisal

The increasing number of RCTs in SDT interventions and reported efficacy has made the evidence base stronger for using SDT as the theoretical framework for developing a psychoeducational intervention for family members at-risk for ICCs. Nevertheless, it is important to be aware that there is a risk of bias in these studies. Based on the Cochrane Risk of Bias tool (Higgins and Green, 2009) the most common is lack of concealment for treatment allocation (Badr et al., 2015, Cossette et al., 2016, Duda et al., 2014, Dunbar et al., 2013, Edmunds et al., 2008, Mayer et al., 2018, Shah et al., 2016, Stamp et al., 2016), which could be argued as impractical considering the nature of the interventions; and lack of blinding for the outcome assessor (Badr et al., 2015, Duda et al., 2015, Duda et al., 2014, Dunbar et al., 2013, Edmunds et al., 2008, Shah et al., 2016, Williams et al., 2006).

Samples sizes were also relatively modest. Despite these methodological issues, SDT is very much at the forefront of providing empirical evidence as the path analyses conducted on the SDT models within the RCTs provides an important insight into causality in which is often missing in behavioural change research (Michie et al., 2016).

4.5 Conclusion and recommendation

Overall, the congruence of SDT to the needs of family members at-risk for ICCs identified in the systematic review, the growing evidence for efficacy of SDT-orientated interventions in the health domain; and upholding of the theoretical assumptions of SDT

in the path analyses has justified the appropriateness of SDT for use as a theoretical framework in the development of the psychoeducational intervention in this research. This research was also an opportunity to improve on the body of work in SDT by considering the strengths and weaknesses in the previous SDT intervention and trial designs and applying this to a new field in the health domain.

Chapter 5: Phase 2 Developing the intervention model – Qualitative study aims and methods

5.1 Introduction

In this chapter, the qualitative study involving family members who have undergone cardiac screening and/or PGT, Phase 2 of developing the intervention, is reported. This chapter provides the background and justification for the qualitative study (Section 5.2); as well as the aim (Section 5.3) and the methods used (Section 5.4). This chapter concludes in Section 5.6, providing a lead into the next chapter reporting the findings of the qualitative study.

5.2 Background and justification

The systematic review in Chapter 3 has yielded the preliminary evidence base for the psychoeducational intervention by highlighting the needs of family members at-risk for ICCs. Whilst the review gave a comprehensive and evolving account of the experiences of family members as they underwent screening and/or PGT, it did not explicitly describe the form and manner by which a psychoeducational intervention should be delivered. Furthermore, the studies included did not look at the role of other health professionals involved in the care of ICC patients, such as the specialist nurse, who tends to be the main contact for patients long-term within the specialist ICC clinic; and how they impact on the patient experience. With the mainstreaming of many genetic conditions into disease-specific clinics, rather than remaining in clinical genetics, nurses are increasingly relied on to provide genetic healthcare and counselling (Kirk et al., 2014, Torrance et al., 2006).

The application of Self-determination theory as the theoretical framework for this research is a novel approach in the field of ICCs and whilst a valid justification was given in Chapter 4, a primary study utilising SDT in this population will support applicability. Therefore, in accordance with the development phase of the MRC Framework for the

development of complex interventions, this qualitative study will add to the findings of the systematic review to generate a comprehensive intervention model as well as provide the feedback loop to strengthen the evidence base for the intervention (Bleijenberg et al., 2018a).

5.3 Aim of the study

The aim of the qualitative study was to obtain the perspective of the intended end-users to inform the content and features of the psychoeducational intervention being developed for this research. By conducting participatory group discussions or interviews with family members who have undergone cardiac screening and/or PGT, their views, preferences and experiences brought forward the content, design, and delivery aspects of the intervention within a contemporary clinical context.

5.4 Method

5.4.1 Study design

Qualitative research attempts to make sense of or interpret a phenomena according to the meanings participants attribute to them (Denzin and Lincoln, 2011). A qualitative research design was chosen for this study as it is most pertinent in providing an in-depth, interpreted understanding of the social milieu of family members at-risk for ICCs and how they make sense of this (Ritchie et al., 2014). Due to the exploratory work remaining to complete the Development stage of the MRC Complex intervention framework, a qualitative approach allowed a greater understanding of the perceptions of family members at-risk for ICCs on how they navigated their way through their screening and/or PGT pathway, the factors that influenced their decision making and adjustment to their results; and their preferences for any psychoeducational intervention to be developed to support them.

Designing qualitative research is dependent on the purpose and objectives of the study, as well as its context (Ritchie et al., 2014). The specific methodological choice was also guided by the worldview or overarching philosophy of pragmatism where mixed methods are adopted to ensure the appropriate methods are chosen to answer the research question rather than focusing on the methods themselves (Cresswell and Plano Clark, 2017). In this phase of the project, to achieve the aims and objectives, a subjective approach by interacting with research participants was required.

5.4.2 Research ethics

This Phase 2 study was given ethical approval by the London-Fulham Research Ethics Committee (REC) on January 16, 2017 (reference: 17/LO/0059). Permission to conduct the study was granted by the NHS Health Research Authority on January 31, 2017. The study poster, participant information sheet, consent form and topic guide can be found in Appendix 2.

5.4.3 Study setting

This study took place within the specialist adult ICC clinics at two inner London centres. These two centres combined make it one of the largest service providers for ICCs in Europe with over 5,000 patients seen each year and therefore caters to a diverse population in terms of ICC conditions, socio-cultural and economic backgrounds. These centres provide the full array of services as per international guidelines for ICCs using a multi-disciplinary team approach and therefore represents the standard of clinical care for patients and families. By virtue of these features, the setting provides an appropriate context for the conduct of the study and should bring about insights which could be applied to the wider population of family members at-risk for ICCs.

5.4.4 Study participants

The following eligibility criteria for entry into the study were guided by the aims of the research and in consultation with the King's Health Partners Patient & Public Involvement for inherited Cardiac Conditions (KHP-PPIICC) group:

- Patients who were aged 16 and older
- Patients who were at-risk relatives who have undergone cardiac screening and/or PGT for ICCs
- Patients who were at-risk relatives who were referred for cardiac screening and/or PGT for ICCs but declined these tests

Index patients (probands) who were attending clinic were not recruited as they were not the target population of the intervention being developed. Those who were under 16 years of age were excluded from the study as they were not within the remit of the adult ICC clinic and in a real-world setting would require tailored interventions for their age group with its accompanying safety governance.

It was considered whether it was possible to recruit patients who had insufficient command of written and spoken English, however, translation services for written materials and individual interviews or focus group discussions were beyond the scope of resources for this study. Furthermore, translation may not capture the ideas, concepts, and feelings as intended and therefore not reach cultural equivalence and congruence of values in the language used (Regmi et al., 2010). This is of vital importance to capture the nuances of the language used, particularly in eliciting SDT concepts.

5.4.5 Sample size

In qualitative research, determining the sample size is not based on producing statistical estimates of the prevalence or distribution of a characteristic to enable application to a wider population. Rather than focusing on the number of participants, the basis of an adequate sample is how much their features are representative of the sampled

population (Ritchie et al., 2014). Whilst the sampling strategy discussed in the next session was designed to this end, ethics committees required an estimate of the number of participants to be recruited. Based on published guidance (Krueger, 2009), PPI input and feedback from peer review of this research, it was agreed that a total of 24 participants, participating in either group discussions or semi-structured interviews would be sufficient to achieve a wide coverage of the patient experience.

5.4.6 Sampling method

Qualitative research generally uses non-probabilistic methods for selecting the sample for the study. The rigour and precision stems not from having a statistically representative sample but rather one that comprehensively represents the salient characteristics of the population (Ritchie et al., 2014). This study therefore employed purposive sampling using the eligibility criteria in Section in 5.4.4 to ensure a thorough exploration and understanding of the perspectives of the target population for the intervention (Bryman, 2012). Although patients would have had the shared experience of undergoing cardiac screening and/or PGT, there are a variety of ICCs, patients differed in terms of the antecedents of their referral into the clinic; and there are different outcomes to screening and/or PGT. Therefore, the maximum variation sampling approach was taken to ensure the perspectives from these varied backgrounds are captured; and to identify cross-cutting themes across this diverse group (Bryman, 2012, Creswell, 2013).

5.4.7 Sampling and recruitment of participants

5.4.7.1 Patient identification

A member of the clinical team, usually the Cardiac genetics nurse, identified eligible patients from their ICC clinic list for the day based on the study inclusion/exclusion criteria. The clinical team member asked the patient if they were willing to be introduced to me, the researcher. The potential participant was then referred to me then given a participant information sheet (Appendix 2 B) to read as they waited for their appointment.

After their clinic appointment, if patients wished to join the study, they were directed by the clinical team to me for an in-depth conversation about the study and an opportunity to ask any questions. It was made clear that they had the option of joining a group discussion or have an individual interview. If they indicated that they wished to participate, patients were screened for eligibility. If they were not eligible, they were thanked for their time and no personal details were recorded.

Eligible patients were asked for their contact details which was recorded in the Patient Identification Log. At least 48 hours were given to patients to fully consider joining the study and to provide the opportunity to discuss this with their family or significant others or a member of their clinical team. Follow-up phone calls were made to verify the patient's interest in participating and understanding of the study; and to address any questions or concerns.

Purposive sampling facilitates a wider coverage of the sample therefore it is important to consider the subgroups within the study population; and work methodically in the final selection of the sample (Silverman, 2013). Certain situations dictated the resulting sample matrix and the composition of the groups during data collection; this was associated with the groups of patients attending the ICC clinic during the recruitment period. Firstly, to provide a wider perspective, patients who declined screening and/or PGT were included in the proposed sample, however, those who attended clinic during the recruitment period tended to proceed with tests. In this aspect, the specialist nurse was able to identify patients from their caseload who met the inclusion criteria and put them forward for recruitment. Secondly, as it is the practice in the recruitment sites that cardiac screening for family members is initiated soon after a proband is given a clinical diagnosis, this did not allow for a situation where relatives must wait for the results of the proband's genetic test (which paves the way for their PGT) before attending the clinic. Therefore, most patients who have undergone PGT would have had a prior cardiac screen.

Finally, ICC clinics are not set up as family clinics per se, but family members can request to attend on the same day. During the recruitment period, some family members attended together and although they were recruited individually, when preference for a group discussion was raised, all family groups, except one husband and wife couple, decided they would like to be interviewed as a group in their home. This resulted in seven family group interviews, two mixed (non-family) group interviews having only 2-3 participants, three individual interviews; and a larger sample size (N=29). Despite these circumstances, the resulting sample matrix (Table 5) represented a varied range of relevant experiences.

Table 5 Sample matrix for at-risk family members undergoing cardiac screening and/or predictive genetic testing for inherited cardiac conditions

Family history	Cardiac screening only			PGT only		Both		Declined	Total
	Pos	Neg	Uncertain	Pos	Neg	Pos	Neg		
HCM		6	2			2	3		13
DCM	3	2							5
ARVC			1			3	1		5
LQTS				1		2	1		4
Brugada						1		1	2
Total	14			1		13		1	29

Abbreviations: Arrhythmogenic Right Ventricular Cardiomyopathy-ARVC, Dilated Cardiomyopathy-DCM, Hypertrophic Cardiomyopathy-HCM, Long QY Syndrome (LQTS), Negative-Neg, Positive-Pos, Predictive Genetic Test (PGT)

5.4.7.2 Consent

If patients confirmed they would like to participate, they were asked to sign the patient consent form (Appendix 2 C) which was sent via post. The patient placed their initials at the end of each statement in the consent form indicating their agreement and affixed their signature and date at the end of the form next to their printed name. Upon receipt of the consent form, the participants could contact the researcher at any time if they wanted more information about the study. The signed consent form was returned in a pre-paid envelope, and the researcher would also sign the patient consent form. The original form was kept securely, and one copy filed in the medical notes and another copy was retained by the patient. An alternative arrangement often preferred by the participant was to go over the consent form on the day of the group discussion or interview and affix their signature prior to data collection.

Participants could withdraw from the study at any time without requiring a reason and it was emphasised that their clinical care would not be compromised. It was made clear in the patient information sheet and in the consent form that no further data would be collected from them. Any data already collected would be used in the data analysis but will be anonymised.

5.4.8 Data collection

Data were collected via group discussions or in-depth, individual semi-structured interviews. Participation in a group discussion was the initial option offered to the participant as this would allow interaction amongst the group and generate useful qualitative data from responding to the researcher's questions as well as spontaneous dialogue with each other (McLeod, 2011). It was recognised that some of the topics surrounding ICCs could be sensitive and therefore to maximise participation of those

who felt they could not discuss this in a group, a semi-structured interview was also given as an option.

5.4.8.1 Topic guide development

In contrast to survey studies where a pre-defined, fixed set of questions are used, qualitative interview studies tend to be conducted with smaller groups of people and have a more informal and naturalistic pattern of questioning, allowing the interviewee to set the pace (Silverman, 2013). However, it is recommended that a topic guide is available as an aide-memoir of what should be explored during data collection and can support the researcher to manage an unpredictable social situation (Ritchie et al., 2014).

The topic guide (Appendix 2 D) for this research was developed based on the synthesis in the systematic review in Chapter 3 and principles of Self-determination theory, as well as the clinical expertise of the researcher. The topic guide was reviewed and agreed on by the KHP-PPICC group and academic supervisors. The points of discussion were organised to begin with contextual information then onto unthreatening topics to ease introductions across the group before moving on to the main topics. The topics explored included the participants' understanding of their genetic risk and perception of their current health status, how they experienced cardiac screening and/or PGT and the salient aspects of that care; and what support they received or would have liked to receive focusing on the three basic psychological needs-competence, autonomy, and relatedness.

5.4.8.2 Location and set up

The decision for the location of the group and individual interviews were made based on maximising patient participation and minimising any associated burdens. Therefore, the needs of participants for convenience, comfort and privacy were considered. The group and individual interviews were held in a booked room on the University premises which

was easily accessible by public transport, or in a patient's residence, depending on their preference and times were flexible. Refreshments were provided and transport and childcare costs were reimbursed.

When the sessions occurred in a participant's home, the researcher acknowledged and thanked other members of the household who were not being interviewed. Participants with children tended to provide toys and activities to occupy them and a room where it was relatively quiet was used. Respect for house rules such as taking shoes off when entering the household were followed.

5.4.8.3 The interview process

All the group sessions and individual interviews were digitally recorded using an encrypted voice recorder with a satellite microphone. Depending on the size of the group and room, the recorder was placed adjacent to the researcher to enable checking for proper functioning during the session; and the microphone more centrally to ensure clarity. The microphones were small and discreet to avoid unnerving the participants with too much equipment (Al-Yateem, 2012). Tissues and water were always available as sensitive issues may be touched on and arrangements had been made with the clinical team in case any referrals to them for clinical care were needed.

5.4.8.3.1 Group interviews

The rationale behind positioning participation in group interviews as the primary mode of data collection was their ability to generate data and insights from both the participants and their interaction. The social context groups provide was consistent with relatedness and autonomy in SDT; and offers the opportunity for a more natural environment where participants influence and are influenced; and shows how collective and self-identity are constructed and expressed (Krueger, 2009). Whilst group sessions should appear fluid

and spontaneous, it is a formal interaction that must be moderated carefully (Puchta and Potter, 2004).

There were several stages in the moderation of the group interviews (Ritchie et al., 2014) which commenced as each participant arrived. In the case of a group interview in the family's home, the family would have typically designated a space where the discussions can take place. The researcher would take a few minutes to work out an ideal position with the host to ensure maximal visibility and interaction with the group. The researcher then welcomed and thanked the participants, offered refreshments, and participants were put at ease with friendly conversation that did not stray into the topic guide. Once all the participants were present, a formal opening of the session was done by the researcher which started with a personal introduction, followed by an explanation of the purpose, an outline of the research, and who funded it. A description of how long the session would last, how it will be recorded and how the data obtained will be used and protected was given. The 'ground rules' in terms of confidentiality, refraining from using mobile phones and respect for others' opinions were agreed on by the group. The importance of each person's views was stressed alongside the voluntary nature of participation and the extent of information they wish to share. This alerted the participants to the possibility of sensitive and personal topics that may be discussed; thus, it was also emphasised that they were free to withdraw from the session at any time without having to give an explanation. It was also explained that any reimbursements would be dealt with at the end of the session. An opportunity for questions or clarifications was given before the recording commenced.

When all the participants confirmed that they would like to proceed with the discussion, individual introductions were made by the participants themselves and they were encouraged to share something about themselves, not necessarily related to the ICC affecting them, such as a hobby or their profession. This helped to build a degree of

familiarity and started practicing the role of that of a speaker and listener, which is required in group dynamics (Ritchie et al., 2014). This second stage was followed by an introduction of the opening topic by the researcher which was a broad inquiry of how 'things were going' in general for each participant to make it easy for verbalisation of spontaneous thoughts. Generally, those who responded with general answers would have follow up questions from the researcher to encourage discussions related to their health and ICCs. This was typically the springboard where participants started spontaneously asking each other questions to compare and contrast their experiences.

The main discussion would flow after these initial stages and the researcher would continue to balance the need to promote the group interaction against making sure an individual perspective is heard, and the value of active debate against covering other topics. Throughout these interactions, the researcher remained flexible to allow participants to introduce topics of interest to them but 'topical steering' was required to make sure discussions remained relevant (Flick, 2009). This was particularly needed when individuals would start asking for clarifications about their own medical concerns, whilst this was acknowledged and noted, the researcher would ask a question related to this concern in the topic guide to draw out how the participants coped and felt about this to return to the purpose of the research. Other techniques used were to use silence to allow the group time to reflect followed by further probing if a point needed to be elaborated or moving on to the next topic. Non-verbal cues and clues were also noted as other ways of participants indicating that they were in agreement/disagreement with what was being said or disengaging with the conversation; therefore, at times, non-verbal communication was needed as a prompt to draw them in to share their views (Ritchie et al., 2014).

An important task the researcher had was to ensure the balance of between individual contributions as self-disclosure required in group discussions can be difficult for some

people whilst others can dominate the conversation (Krueger, 2009). In family groups, where there is high degree of familiarity and established 'key communicators' (Metcalfe et al., 2011) it was also important to bring out the unique opinions other members may hold. When a participant provided an opinion on behalf of the family group, the researcher looked for non-verbal cues from others indicating non-concordance. If this was evident, the researcher would seek clarification and often this allowed for a broader, but not necessarily contradicting view.

Non-verbal communication from the researcher was used in a subtle way to create space for everyone to contribute. This was done by nodding and looking attentively at the person speaking to indicate they have been heard then glancing at the next person as a prompt to join the conversation or casting a wide glance at the group to indicate that the floor is open to a new speaker. Eye contact is very important to encourage and give confidence to a reticent participant. There was no need to address dominant participants verbally as the group appeared to take non-verbal cues from each other and naturally took turns at speaking (Flick, 2009).

To avoid ending the discussion abruptly, a final topic was usually decided midway through the group discussion as it is often difficult to foresee the flow of the conversation. The researcher took mental notes of what has already been covered in the session to enable this relatively quick decision (Puchta and Potter, 2004). The final discussion tended to involve more direct questioning on what the group felt was most useful in supporting them through their screening and/or PGT journey or suggestions for what could be done better. This helped draw out what impacted on their well-being the most and ended the session on a positive and completed note (Silverman, 2013), as well as facilitate prioritisation of features to be developed in a psychoeducational intervention. Finally, the group members were thanked for their participation, the confidentiality of the

information shared was reiterated; and any administration for reimbursements were completed.

5.4.8.3.2 Semi-structured interviews

Participants who did not want to participate in a group interview or could not come to a specific session were given the option to have an in-depth face-to-face interview. This helped in gathering a diverse group of participants to capture the breadth of experiences of family members at-risk for ICCs, particularly those who may have views they felt were radically different from 'the norm' (Marshall and Rossman, 2016). An in-depth interview is a flexible but structured, interactive method to enable an in-depth exploration of a participant's experiences and the meanings they attribute to these. The in-depth nature allows the interviewer to explore the underpinnings of the participants' answers whilst also giving space for reflection by which the participant may generate new knowledge to propose ideas or solutions about a topic (Ritchie et al., 2014).

There are also stages in the semi-structured interview (Robson, 2002), which generally follows those of the group discussions, hence, preparations and procedures were followed in a similar fashion. However, more effort was placed in building rapport and trust at the early stages of the interview to ensure the participant was at ease and to show genuine interest in their story (Patton, 2002). Key to this task was good non-verbal communication such as maintaining eye contact and active listening. Good listening skills were essential to enable the researcher to take note of what has not been said, capture the nuances in participant's account; and to ask relevant follow-up questions (Hammersley and Atkinson, 2007).

In the main stages of the interview, a questioning technique that was clear, open, and non-leading was used to help participants narrate their experiences in depth to bring about thoughts, feelings and emotions that may have been put aside or laid dormant.

Mapping questions were particularly helpful when participants spoke about their experiences and mapped these along the timeline of their cardiac screening and/or PGT. Prompts were offered when a segment of the timeline was not mentioned, or it was not elaborated in full. Gentle probing proved helpful in exploring the impact of events such as a sudden death in the family and in challenging any inconsistencies especially when a set of recommendations was felt to be appropriate for one set of people but not for them, though they appeared to be in the same situation (Ritchie et al., 2014).

Throughout the interview, ample time was given so that the participant could construct and verbalise their answers fully, periods of silence were respected to facilitate reflection (Robson, 2002). Although the topic guide helped ensure that the discussions remained within the focus of the research, talking about parallel subjects such as how participants dealt with other health conditions and other interests were encouraged to get a sense of their wider context. To signal the end of the interview, the researcher would tend to ask if there were any important issues left unmentioned. In a similar fashion to the group discussions, the aim was to end on a positive note with a request for suggestions to improve their experience and those of others yet to undergo screening and/or PGT.

5.4.8.4 Field notes

The use of field notes was an opportunity to record what the researcher saw and heard outside the immediate context of the focus group or interview discussions (Ritchie et al., 2014). The accounts were both descriptive and reflective, including thoughts about the dynamic of the encounter (both in relation to the researcher and the group) and issues relating to the environment or context of the interview. Field notes were completed at the earliest opportunity following the focus group or interview sessions to avoid bias from intervening events. In the research process, these field notes helped the researcher for the next data collection session, whether this was for improving the environment (choosing a room with less traffic noise) or thinking about strategies to avoid veering into

clinical consultations. During data analysis, these field notes proved particularly helpful in contextualising the participants' responses (Silverman, 2013).

5.4.8.5 Data management

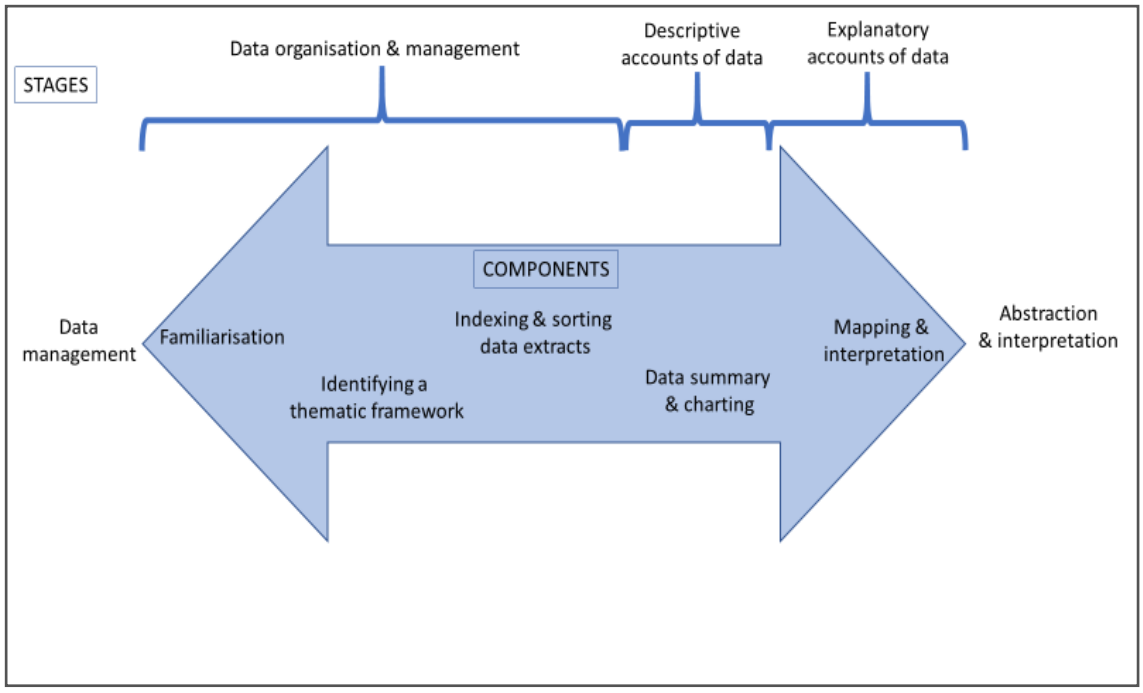
Data management, interpretation and project management are key capabilities of standard Computer-assisted quality data analysis software (CAQDAS) that is widely used in qualitative research. The main benefit of using CAQDAS is the speed with which it can handle large volumes of data. It is also thought to improve rigour of analysis as researchers are able to order, search and filter data systematically; and demonstrate that a search has been conducted across all the data (Flick, 2009). Whilst there are arguments that CAQDAS can be intellectually stifling and inhibits data analysis (McLafferty and Farley, 2006), these programmes have now gone through multiple iterations and they have more flexibility and functionality to adapt to the analysis process (Seale and Rivas, 2012). It is important to note that CAQDAS does not do the analysis and therefore the researcher is central to defining the analytical issues to pursue, identifying the important ideas and how to represent these appropriately (Moser and Korstjens, 2018).

The chosen method of analysis in this study was Framework Analysis which uses framework matrices to organise data by case and code to facilitate data analysis (Ritchie and Spencer, 1994). NVivo® version 11 has the functionality to generate framework matrices therefore it was the CAQDAS chosen for this study. The programme allows a large amount of data to be viewed within cases and across cases thereby preserving its context. Query features can be used across the whole dataset and assist in retrieving quotations that back up the analytical findings (Ritchie et al., 2014). The transcriptions of both the focus group discussions and interviews were entered into NVivo® software.

5.4.9 Data Analysis

In this section, the application of Framework Analysis in this study is described in detail (Ritchie and Spencer, 1994). There are five non-linear, and interlinked components in the formal analysis process (Figure 9). Divided into stages, these components build on each other to achieve levels of interpretation as well as providing the opportunity to look back and reflect on how consistently the original material is being represented (Miles and Huberman, 1994).

Figure 9 The Framework Analysis Process



5.4.9.1 Stage 1 Data organisation and management

This stage encompasses familiarisation with the data, the application of the thematic framework and the indexing of extracts. The familiarisation process involved immersion in the data by initially listening to the recordings whilst simultaneously reading the transcripts to verify accuracy. During this process, each transcript was anonymised to ensure people and places were not identifiable. All the transcripts were read and re-read

alongside the field notes. This continued until it was felt that the diversity of circumstances and features within the data set was understood (Ritchie et al., 2014). Transcripts were assigned a number from 1-12 and data management of each was conducted consecutively. Each participant was assigned a pseudonym which facilitated linkage to the field notes and demographic information while maintaining anonymity. The functionality of NVivo11® of assigning attributes to each participant facilitated connection to subgroups. A full description of participant characteristics can be found in Chapter 6.

The development of codes for each dataset to establish the thematic framework was an iterative process featuring both deductive and inductive techniques. Each transcript was read, and codes were developed from key words, statements, and discussions. Codes were labelled as close as possible to the language found in the recordings. This process was conducted independently by the researcher and one of the supervisors (ER). The codes generated by the researcher and the supervisor were reviewed side by side for consistency, commonalities and differences and resulted in a common initial inventory of 67 descriptive codes. These codes were checked against the findings of the systematic review in Chapter 3, the objectives of the study; and the topic guide to ensure comprehensive coverage and relevance of each code.

Underlying ideas or ‘themes’ that linked particular codes were then identified and codes were grouped and sorted according to different levels of generality. A consensus for the thematic framework for organising the data was reached through collaborative discussions with the supervisory group. This included discourse on the different views on labelling and grouping of codes; and definitions which helped the researcher become aware of biases and blind spots. An excerpt from the thematic framework is presented in Table 6.

Table 6 Excerpts from the thematic framework

Extracts from transcripts	In-vivo codes	Descriptive codes	Initial theme
I think it's frightening considering obviously my dad passed away from the condition. I know he probably had a lot more going on, age and other issues related, slightly different, but yes, frightening really to think, "Oh hang on," then they start looking at you and thinking you now have this inherited condition. And knowing that somebody died of it at such a young age and not enough is known about it and you don't know what you're going to be like in five, ten years' time.	"I think it's frightening" "my dad passed away from the condition. "And knowing that somebody died of it at such a young age" "not enough is known about it and you don't know what you're going to be like in five, ten years' time."	Emotional reaction to result Worries and fears Worries and fears	Living with the results of cardiac screening and/or predictive genetic testing (PGT)
To be honest, I've always been aware, because my dad died when he was only 50, and we knew it was something to do with his heart. Being aware I might have had a heart – because I know heart conditions can be genetic as well, I've always been aware that there might have been a problem with my heart. Not LQT, but I thought I might be susceptible to a heart attack, which I thought was what killed my dad. So, I've always tried to keep myself healthy and things like that.	"I've always been aware, because my dad died when he was only 50, and we knew it was something to do with his heart. Being aware I might have had a heart – because I know heart conditions can be genetic as well," "I've always tried to keep myself healthy and things like that."	Genetic concepts Thoughts and feelings about risk Lifestyle adjustments	Being an at-risk relative Being an at-risk relative Living with the results of cardiac screening and/or PGT

Extracts from transcripts	In-vivo codes	Descriptive codes	Initial theme
it's just an issue with my information. Sometimes you go in and you wait a long time for the appointment and then they don't have the results. You go, "But I had that monitor on six months ago." It is a bit unsatisfactory sometimes in that, because you only have one slot to see a doctor once a year, and you have to make a big decision: "Do I opt for Beta Blockers or do I need to be doing something else?" But the information you need to make the decision is not there, or for the doctor to help you.	"it's just an issue with my information. Sometimes you go in and you wait a long time for the appointment and then they don't have the results." "It is a bit unsatisfactory" "But the information you need to make the decision is not there, or for the doctor to help you."	Process of screening and/or PGT Thoughts and feelings about screening and/or PGT Information	Experience of cardiac screening and/or PGT Living with the results of cardiac screening and/or PGT

5.4.9.2 Stage 2 Descriptive accounts of data

An essential first step in producing a qualitative research account is to describe the data. Within Framework Analysis this involved writing a summary of all coded data within each transcript based on the thematic framework. Working systematically through the data by theme for each participant facilitated deep immersion in the topic and a more refined understanding of the content and variation. Each summary was carefully written to remain close to the participant's own language with minimal interpretation. These were displayed using framework matrices in NVivo® which retained their links to the original transcript. This linkage made it easy to contextualise the data extracts during the analysis process and an example can be seen in Appendix 6.

Following summarisation, the next step was to capture the linkages or 'what is happening' within a theme and sub-themes. To achieve this, summaries were individually coded to capture the range and diversity of views and experiences expressed by the participants, while listing the elements present in the responses and the dimensions that differentiate them. The range of perceptions and experiences within each code were labelled followed by grouping the data that appeared to be about the same thing. Categories were formed after the identification of link between the data extracts.

At the start, each descriptive code was considered as a potential category. When codes appeared to be part of a similar topic, the links between them were identified and from these, the initial categories were developed (Table 7). The categories were refined, through multiple iterations, by eliciting the key dimensions with the synthesised data and generating associations between them. The resulting categories were refined to form initial sub-themes from which themes were developed that reflected the breadth and integration across the participants' accounts. This iterative process involved discussions throughout all stages of the analysis around the exploration and the interpretations of the data between the researcher and the supervisory team. These discussions were an essential mechanism which supported clarity in analytical decisions, enhanced exploration of the dataset and minimised potential researcher bias. The initial categories and subthemes are summarised in Table 7.

Table 7 Initial categories and sub-themes

Initial categories	Initial subthemes
Health status	Perception of health
Other health conditions	
Concepts of inheritance	Genetic concepts
Recalling family history of heart problems	
Comparing disease manifestation	

Initial categories	Initial subthemes
Knowledge gained from screening and/or predictive genetic test (PGT)	
Source of risk information	Becoming an at-risk relative
Concerns for proband, self and family	
Predicting results of screening and/or PGT	
Motivation for screening and/or PGT	
Source of support and information	
Screening and/or PGT as a process	Experience of screening and/or PGT
Roles of and relationships with health care practitioners	
Opinions on screening and/or PGT experience	
Source of support and information for screening and/or PGT	
The proband before and after diagnosis	Story of the proband
Caring for the proband	
Impact of sudden cardiac death or severe illness of the proband	
Initial reactions to the result	Living with the results of the screening and/or PGT results
Psychological adjustments	
Lifestyle modifications	
Ongoing worries and fears	
Source of support and information for living with a diagnosis and/or carrier status	
Concern for the family	Family communication and other social groups
Communicating risk for at-risk family members	
Families supporting each other	
Support from other social groups	
Suggestions for improving screening and/or PGT experience	Improving clinical services
Opinions on additional support needed	
Wider public awareness for inherited cardiac conditions and prevention of SCD	
Seeking agreement or consensus	Within group or family communication
Giving and receiving advice	
Continuing a thread	

5.4.9.3 Stage 3 Explanatory accounts of data

Due to the large amount of data generated by this study, measures were implemented to ensure that participants' accounts were accurately represented, to minimise misinterpretation; and to achieve comprehensive coverage of all the dimensions within the data. This included reflecting on the original data as a whole and at each step of the analytical stage from mapping to interpretation; coupled with regular discussions with the supervisory team.

To generate explanatory accounts of the data, there was a shift from descriptions of individual cases towards the development of themes which offered possible explanations for what was happening within the data. Themes were generated from the data set by reviewing the matrix and making connections within and between participant and categories. This process was influenced both by the original research aims, the application of Self-determination Theory, and by new concepts generated inductively from the data. This combined approach to data analysis is possible due to the flexibility of Framework approach (Ritchie et al., 2014) and appropriate for ensuring that any unexpected aspects of the participants' experiences are accounted for whilst analysing data within an existing theoretical framework (Gale et al., 2013).

To ensure that findings were not 'forced' to fit preconceived ideas, the early stages of analysis kept as close to the participants' own language and accounts; and SDT concepts were introduced much later on in the analytic process in as far as they actually matched the data (Morrell et al., 2011). This mapping process was regularly checked with the supervisory team to ensure that the explanatory accounts reflected the uniqueness and diversity of the evidence. These interpretations are presented in Table 8 and presented in Chapter 6 which detail the participants' accounts.

Table 8 Subthemes and final themes

Links between initial themes and categories	Subthemes	Final themes
Perception of health Genetic concepts Becoming an at-risk relative Story of the proband	The imprint of sudden cardiac death on inherited cardiac condition (ICC) families *Taking a hit for the team*-from disease comes prevention Towards balanced parenting in ICCs	Impact of the proband's story
Genetic concepts Becoming an at-risk relative Experience of screening and/or predictive genetic testing (PGT) Family communication and other social groups	The pathway to screening and/or PGT process Screening and/or PGT happens-ready or not Through the generations- concerns and actions for other at-risk family members	Leveraged autonomy
Perception of health Genetic concepts Experience of screening and/or PGT Living with the results of the screening and/or PGT results Improving clinical services	Baseline information, building competence Communication with health professionals-from the straightforward to mixed messages Participants who had a normal screen or non-genetic carrier results-going back to normal Participants with a possible or definitive ICC diagnosis and/or genetic carriers-*getting on with it* in hope Variable penetrance in ICCs requires tailored health advice Accessing psychological support-*It's a good idea for some*	Harnessing competence
Experience of screening and/or PGT Family communication and other social groups Improving clinical services Within group or family communication	Home is where heart education is-coaching within families The clinical service and the health professionals in ICCs-*scoops you up* The listening ear of friends, employers, and patient support groups	Relatedness in the social context of ICCs

5.4.10 Study trustworthiness

There have been multiple debates and suggestions for standards by which the rigour of qualitative research should be evaluated (Polit and Beck, 2006, Rolfe, 2006, Stige et al., 2009), however, it is generally accepted that criteria used for quantitative research are not appropriate or adequate (Korstjens and Moser, 2018). Sound and explicit descriptions alongside rich and innovative interpretations are hallmarks for rigour in qualitative research (Polit and Beck, 2006). In the pursuit of methodological rigour and trustworthiness, the researcher used the evaluative framework proposed by Lincoln and Guba (1985) to illustrate the provisions made to achieve this. This framework helped elicit the credibility of the presentation and interpretation of the data; transferability to other settings and dependability of the findings; and confirmability, which demonstrated the congruence of interpretation of the data between researchers (Lincoln and Guba, 1985, Shenton, 2004, Treharne and Riggs, 2015).

With reference to Lincoln and Guba's (1985) framework, the measures taken to enhance rigour and trustworthiness of this research include:

Credibility

Regular meetings to discuss analytical decisions provided opportunities to keep biases in check, consider blind spots; and to assess interpretations emerging from the data. Deviant cases were considered and included in the explanatory accounts to capture the breadth and range of patterns in the data.

Framework Analysis facilitated the retention of proximity to the original data, retaining the context of the data extracts in the explanatory accounts.

Transferability

Purposive sampling of participants was deployed, incorporating a diverse range of experiences in family members at-risk for ICCs.

The participants' characteristics were accounted for in detail, with provisions for anonymity, which provided context to their contributions.

The research sites were described in adequate detail with attention to minimising risk of identifying participants alongside a full description of the data collection tools and methods.

Dependability

To check for dependability, anonymised preliminary findings of the data were shared with the patient and public involvement group comprised of members who had similar backgrounds with the participants, but who were not involved with the study. Initial interpretations as well as the final representations of the participants' accounts, as presented in the following chapters, were discussed as part of the process of modelling the intervention.

Confirmability

To ensure confirmability, the researcher practiced reflexivity to bring out personal predispositions. These personal biases were acknowledged and addressed within the analytical discourse at all stages, with support from the supervisory group.

A record of how analytical decisions were made with examples of data management and analysis were presented (Sections 5.4.8.5-5.4.9.3).

In presenting the study findings, these were supported with direct quotes from the participants.

5.4.11 Reflexivity

The consistent practice of reflexivity was of vital importance in conducting this research as this process of critical self-reflection helped identify personal biases, preconceptions, and preferences, as well as bring about aspects of the researcher relationship with the participants; and how this affects the way they respond to the researcher (Polit and Beck, 2017). Reflexivity highlights that the researcher is not a passive participant, and therefore must continuously reflect on how they actively shape the study (Holloway and Galvin, 2016).

My clinical background and role were an integral part of the motivation for undertaking this research as detailed in Chapter 1. The most challenging aspect of conducting this research was trying to delineate my role as a nurse researcher and my identity as a clinical nurse specialist. Whilst I was continuously mindful of this at every step of the research process, at times, I was also challenged by other researchers and my supervisors.

This balancing of clinical versus research roles was critical during data collection as participants knew of my background and the interviews and group conversations would sometimes stray into the clinical realm. Apart from an awareness of the aspects of care in ICCs, there were some instances where I may have looked after a member of their family some time ago which brought about familiarity and a risk of making assumptions which could have curtailed the depth of the conversations or made participants hold back on the range of opinions they have about their care. I was conscious of this potential bias, and therefore communicated my researcher role at the outset, endeavoured to save

general clinical queries towards the end of the sessions; and any specific queries were directed towards their clinical team.

The strategies employed to support reflexivity included keeping a reflective diary/field notes from each participant contact, writing of memos during data management and analysis; and the review of data transcripts by the supervisory group to check the flow and coverage of the interviews or focus group discussions. Regular sessions with the supervisory group to help focus my lens as a researcher, particularly during the data management and analysis stage, helped enable a more robust interpretation and minimised interplay with personal biases and preconceptions.

5.5 Summary

The justification and aims of Phase 2 of this project were outlined in this chapter. The methods by which the qualitative study was conducted, the findings of which were a key component of developing the PISICC intervention model, were discussed in detail. This included describing the analytical steps taken utilising Framework Analysis underpinned by Self Determination theory with examples of progression from data management, description to interpretation. In the next chapters, the findings of this study are reported followed by a synthesis of evidence gained from the two phases of the PISICC project to arrive at a proposed intervention model

Chapter 6: Phase 2 Developing the intervention model – Findings from the Qualitative study

6.1 Introduction

This Findings chapter is divided into two parts: The characteristics of the participants are described in the first part and then in the second part, the analysis of the accounts of the at-risk family members undergoing cardiac screening and/or PGT for an ICC are presented thematically.

6.2 Participant characteristics

These findings were derived from a total of nine group interviews and three individual interviews which were conducted with 29 participants from 14 families, who were purposively sampled as previously described. Table 9 summarises the participant characteristics and are grouped according to families where applicable. Seven out of the nine group interviews were comprised exclusively of participants of the same family and one group consisted of a husband-and-wife couple and one unrelated participant; and another group was comprised of two unrelated participants. There was a total of 17 women (59%) and 12 men (42%), with an age range of 18-83 y/o. Most of the participants were White British (93%) and two participants were British Asian.

Participants had a varied ICC family history with HCM (n=14) predominating, followed by DCM (n=5), ARVC (n=4), LQTS (n=4) and Brugada (n=2) and were either parents (n=9), siblings (n=14) or adult children (n=6) of probands. The proband's clinical course ranged from severe-cardiac arrest (n=3, 1 family), death (n=13, 7 families), moderate (n=12, 6 families) to mild (n=1, 1 family).

Most participants either had cardiac screening only (48%) or had both cardiac screening and PGT (45%). There was one patient who had PGT only for LQTS and one patient

who declined cardiac screening. For those participants who only had a cardiac screen, three were diagnosed with an ICC while another three had unclear results, most had normal screens (57%, n=8). For participants who had both cardiac screening and PGT and PGT only, most (64%, n=9) were found to be genetic carriers for an ICC. Due to the variable penetrance in ICCs, it is expected that those diagnosed with an ICC and/or are genetic carriers may or may not manifest symptoms therefore it is not surprising that most participants were asymptomatic (66%, n=19). Of the three participants with unclear screening results, one was asymptomatic, another symptomatic and the other had symptoms for another health condition. Some participants who had negative screens or genetic tests had other signs and symptoms for other health issues (n=4) whilst one participant with uncertain screening results also had signs and symptoms for a different health issue. The lone participant who declined screening did not exhibit any signs and symptoms at the time he was invited for cardiac tests. However, by the time he participated in this study, he revealed that he had needed to seek medical attention at another centre due to dizzy spells which subsequently resulted in a diagnosis of Brugada Syndrome.

The range of the time period from the receipt of their cardiac screening and/or PGT result was 0-6 months to over 6 years. Rather than assigning non-specific codes, participants were assigned a pseudonym to preserve their anonymity and when quoted, will be followed by family number (FX) (if applicable), the ICC history and cardiac screening or PGT status.

Table 9 Participant Characteristics and Demographics

Family Number or Individual	Name	Sex	Age Group	Ethnicity	Occupation	Family Diagnosis	Relationship to proband	Proband history	Test	Result	Health Status	Time since test result
1	Fred	Male	55-64	White	Professional	LQTS	Parent	Severe-cardiac arrest	Both	Positive screen & carrier	Asymptomatic	4-5 years
	Julie	Female	35-44	White	Professional	LQTS	Sibling	Severe-cardiac arrest	Both	Positive screen & carrier	Asymptomatic	4-5 years
	Val	Female	55-64	White	Professional	LQTS	Parent	Severe-cardiac arrest	Both	Negative screen & non-carrier	Asymptomatic	4-5 years
2	Bob	Male	45-54	White	Manual	HCM	Sibling	Moderate-controlled with meds or devices	Cardiac screening	Unclear	Asymptomatic-other co-morbidity	2-3 years
	Ken	Male	55-64	White	Manual	HCM	Sibling	Moderate-controlled with meds or devices	Cardiac screening	Negative screen	Asymptomatic	2-3 years
	Sheila	Female	over 75	White	Retired	HCM	Parent	Moderate-controlled with meds or devices	Cardiac screening	Negative screen	Asymptomatic-other co-morbidity	over 6 years
3	Jo	Male	35-44	Asian	Manual	ARVC	Child	Death	Both	Positive screen & carrier	Symptomatic	over 6 years

Family Number or Individual	Name	Sex	Age Group	Ethnicity	Occupation	Family Diagnosis	Relationship to proband	Proband history	Test	Result	Health Status	Time since test result
	Pam	Female	35-44	Asian	Professional	ARVC	Child	Death	Both	Positive screen & carrier	Symptomatic	over 6 years
4	Bill	Male	35-44	White	Manual	DCM	Sibling	Death	Cardiac screening	Positive screen	Asymptomatic	0-6 months
	Jane	Female	25-34	White	Professional	DCM	Sibling	Death	Cardiac screening	Positive screen	Symptomatic	7 months-1 year
	Linda	Female	55-64	White	Professional	DCM	Parent	Death	Cardiac screening	Positive screen	Asymptomatic	2-3 years
	Tina	Female	35-44	White	Professional	DCM	Sibling	Death	Cardiac screening	Negative screen	Asymptomatic	7 months-1 year
5	Louise	Female	25-34	White	Manual	HCM	Sibling	Death	Cardiac screening	Negative screen	Asymptomatic	over 6 years
	May	Female	45-54	White	Professional	HCM	Parent	Death	Cardiac screening	Negative screen	Asymptomatic-other co-morbidity	over 6 years
	Tom	Male	18-24	White	Manual	HCM	Sibling	Death	Cardiac screening	Negative screen	Asymptomatic	2-3 years
6 (interviewed with Lisa)	Pat	Female	35-44	White	Professional	ARVC	Parent	Moderate-controlled with meds or devices	Both	Positive screen & carrier	Symptomatic	7 months-1 year

Family Number or Individual	Name	Sex	Age Group	Ethnicity	Occupation	Family Diagnosis	Relationship to proband	Proband history	Test	Result	Health Status	Time since test result
	Paul	Male	35-44	White	Professional	ARVC	Parent	Moderate-controlled with meds or devices	Both	Negative screen & non-carrier	Asymptomatic-other co-morbidity	7 months-1 year
7	Kate	Female	18-24	White	Professional	HCM	Sibling	Moderate-controlled with meds or devices	Both	Negative screen & non-carrier	Asymptomatic	0-6 months
	Mary	Female	55-64	White	Professional	HCM	Parent	Moderate-controlled with meds or devices	Both	Negative screen & non-carrier	Asymptomatic	2-3 years
	Sharon	Female	18-24	White	Student	HCM	Sibling	Moderate-controlled with meds or devices	Both	Carrier	Asymptomatic	0-6 months
	Steve	Male	55-64	White	Professional	HCM	Parent	Moderate-controlled with meds or devices	Both	Negative screen & non-carrier	Asymptomatic	2-3 years
	Tim	Male	18-24	White	Student	HCM	Sibling	Moderate-controlled with meds or devices	Both	Negative screen & non-carrier	Asymptomatic	0-6 months
Individual (Interviewed with Family 6)	Lisa	Female	25-34	White	Manual	Brugada	Child	Death	Both	Positive screen & carrier	Asymptomatic	4-5 years

Family Number or Individual	Name	Sex	Age Group	Ethnicity	Occupation	Family Diagnosis	Relationship to proband	Proband history	Test	Result	Health Status	Time since test result
Individual (Interviewed w/ John)	Jess	Female	55-64	White	Professional	HCM	Child	Mild-hardly any symptoms	Cardiac screening	Negative screen	Asymptomatic	0-6 months
Individual (Interviewed w/ Jess)	John	Male	65-74	White	Professional	HCM	Sibling	Moderate-controlled with meds or devices	Cardiac screening	Unclear	Asymptomatic	4-5 years
Individual	Maria	Female	35-44	White	Professional	DCM	Child	Death	Cardiac screening	Negative screen	Asymptomatic-other co-morbidity	4-5 years
Individual	Karen	Female	45-54	White	Professional	LQTS	Sibling	Moderate-controlled with meds or devices	Predictive testing	Carrier	Asymptomatic	4-5 years
Individual	Ben	Male	35-44	White	Professional	ARVC	Sibling	Death	Cardiac screening	Unclear	Symptomatic	2-3 years
Individual	James	Male	25-34	White	Professional	Brugada	Child	Death	Declined	N/A	Asymptomatic	N/A

6.3 Analysis of the accounts of the at-risk family members undergoing cardiac screening and/or predictive genetic testing for an inherited cardiac condition

The views, experiences and preferences of at-risk family members undergoing cardiac screening and/or PGT for an ICC were expressed in four main themes: Impact of the proband's story, Leveraged autonomy, Harnessing competence and Relatedness in the social context of ICCs. These are set out in detail in the following sections and are supported with relevant extracts from the interview data. Each theme is comprised of several sub-themes, which collectively construct and represent the accounts of the participants.

In analysing the qualitative data, points which were consistent with SDT concepts and references to contemporary literature are made to support the applicability of SDT as a theoretical framework and elicit the wider context in ICCs and family care.

6.3.1 Theme 1: Impact of the proband's story

The story of the proband in the family was not only the root of how the participants became involved in the ICC service, but also emerged as the cornerstone of actions taken, reactions and emotions felt in the context of risk for an ICC. Whilst talking about the proband was not the focus of the interviews, the participants frequently referred to their relative and gave rich descriptions on the proband's personal qualities, health events, the ups and downs experienced and the support they gave, plan to give or limitations in their capacity as a carer. It is important to note that for those who experienced a sudden cardiac death in the family, talking together about the person who died was important for participants; stories of loss and grief are not often shared publicly as this is sometimes deemed socially unacceptable (Mayer et al., 2013). In this study, participants were very open about their loss and appreciated having an

opportunity to talk together where the group comprised of family members about their individual, and collective, bereavement experiences.

Whether the proband had a benign course, moderate disease controlled by medications and/or devices or a severe ICC leading to a cardiac arrest or sudden death; considerable time was allocated by the participants on reflecting on their relationships with them. None of the participants expressed any lasting difficulties in these relationships. However, there was one participant who described the proband in his family as an estranged half-brother but their communication with each other improved over time. How participants were impacted by the story of the proband can be further described within the three sub-themes as these give greater insight into the those who have a family history of sudden death, the perception of the events of the proband preventing further serious disease in the family; and the profound impact on parents and parenting in those whose children are probands.

6.3.1.1 The imprint of sudden cardiac death on inherited cardiac condition families

This sub-theme conveyed how a death in the family does not just affect individual emotions and actions but also impacts family dynamics (Mayer et al., 2013). The death of the proband was by far the most profound aspect of an ICC in families whether the proband died young or in their later years. This mother of a proband who died of DCM in her 20s puts it plainly:

> *I don't think, for me, anything could have been worse than what happened to my daughter. Nothing. (Linda, F4, DCM, pos screen)*

Grief is a normal emotional response to the loss of a loved one. Individuals will grieve differently, and while there is no single pattern, many will go through disbelief,

yearning, anger, sadness and acceptance (Worden, 2018). Typically a lack of understanding around the causes and circumstances of a family member's death tends to limit the ability of the surviving relatives to make sense of their loss (Harrington and Sprowl, 2012) and therefore supports Bill's (F4, DCM, pos screen) view that the confirmation of the cause of the sudden in his sister helped the family cope better. However, acceptance of the death appeared to be an ongoing process as participants continue to look back, searching for clues on why this happened, intertwined with the character of the proband as illustrated by this exchange from Bill's family members:

> *I was trying to think back and think "Did she have any symptoms?" I was remembering things like what I said about the gym when she said, "Oh, I feel like I'm going to have a heart attack," and stuff like that. (Jane F4, DCM, pos screen)*
>
> *But she was a strong person as well. (Tina F4, DCM, neg screen)*
>
> *She wouldn't tell you if she had a problem really, would she? She just got on with things and wouldn't make a fuss. (Linda, F4, DCM, pos screen)*

In a family where the death occurred five years ago, anger and frustration continued because there were clues, but the ICC was not diagnosed and treated promptly:

> *That was frustrating because for years she'd been living with it and there were signs. Actually, my other sister was here the other day and she was pretty angry about it, she was saying even now, because she (my deceased sister) was going to the doctors with swollen ankles, which is obviously a big sign. (Ben, ARVC, unclear screen)*

The participants felt it was important to be informed of the possibility of sudden death, no matter how reassuring the risk stratification was for the proband. One family, where the proband died suddenly aged 18 with a known ICC diagnosis, wished that the medical team had been more explicit that sudden death could still occur despite being classified as low risk. This undermined their trust in the reassurances that doctors gave them for their own health.

> *And it's not until something as drastic as, like you say, Mike dying, that you realise, oh this could happen and I mean when Mike was diagnosed it was like, "Okay, he's got this diagnosis, okay, he can't do this, he's not supposed to do that, right, fine, but he'll still carry on." (May F5, HCM, neg screen)*

> *But the doctor said, "It's not life threatening, it's not going to kill you," and it did. So that's my biggest worry is like when they say to mum about her heart condition, and I'm like, "No, don't believe them." The same with Mike and he died, and I'm like, "No, I don't trust them if they say it's not life threatening or it's not this," it could be. You just don't know. (Louise, F5, HCM, neg screen)*

This is a new need identified through this study. Whilst there is a great deal of work on estimating the risk of sudden death (O'Mahony et al., 2018), studies on the best practice to convey this specific risk has not been found in the literature. Clinicians appear to be more explicit in conveying the risk for sudden cardiac death when it is higher and tend to downplay it when the risk is low. Yet, despite the knowledge of a higher risk, the sudden death of a family member still takes an emotional toll that requires time to process:

> *Once I knew that he had the diagnosis I knew that that was the risk, in a sense. That he would have a sudden, unexpected death related to heart failure. So although it was obviously a very*

shocking, traumatic event that took a lot of time to come to terms with, it wasn't surprising. (James, BrS, declined screening)

Although the period of profound grief may have passed, the feeling of bereavement and loss was long lasting despite many years after the event. An unexpected death radically changes family roles and tasks. Surviving family members are challenged to live in a world where established family routines and habits that have previously provided a sense of safety and purpose for the family unit is radically disrupted (Neimeyer and Sands, 2011). As a result, participants spoke of how the sudden death either made their relationships closer, break apart or cause them to withdraw from normal life.

It definitely pulled us together. And even now, we still feel like we're still going through the mourning process; it's still a massive loss. My mum has obviously taken it really, really badly. She often cries and yes, a little bit of denial…we're not the most open of families, or at least we haven't been, but I think this has helped us do that. (Ben, ARVC, unclear screen)

Yes, I mean it's made us closer with me and the kids, but obviously with my husband at the time, it actually sort of pulled us apart...we separated two years after Mike died. So, he basically thought I should have been over it by now. (May, F5, HCM, neg screen)

Mothers of sudden cardiac death victims tend to report increased anxiety and depression (Yeates et al., 2013) and although this is not explicitly mentioned by Ben in describing his mother's grief and by May, these could be part and parcel of their prolonged mourning. Posttraumatic stress has also been shown in 44% of first-degree relatives following a young sudden cardiac death (Ingles et al., 2016) and a sign of this is touched on in Tom's account of withdrawing from his normal routines and social groups:

> *I don't know, I think I lost a lot of confidence after he died and I didn't really get it back to be honest. I got really quiet and stuff, in school and everything like that. (Tom, F5, HCM, neg screen)*

Families emphasised that their deceased relative was still present in their lives. Indeed, some expressed that they thought about their deceased relative every day (Ben). Participants made efforts to reconcile themselves to the loss and at the same time reconstructing a life in which the deceased loved one may not be present physically but always with them in spirit (Mayer et al., 2013). Key to keeping the memories of their loved ones alive and to cope with the loss was to think about adopting their ways and attitudes towards life, which has been described as one of the most common forms of posttraumatic growth following bereavement (Calhoun and Tedeschi, 2001) as opposed to being 'stuck' (Ingles et al., 2016) as evidenced in the resolve of this proband's mother & sister.

> *It never stopped him doing anything he wanted to, and I think that's the philosophy that we've adopted as well isn't it? (May, F5, HCM, neg screen)*

> *I live it better…It's more, because Mike was so young when he died, it makes you look at your life and just think, you literally have one life, just live it. Obviously not on the edge, but yes it makes me realise how your life can be so short. (Louise, F5, HCM, neg screen)*

Adaptation to the loss also involved gaining perspective and acceptance of the screening or management regimes that were recommended after a relative's death due to an ICC. The sudden death was a constant reminder that this was the worst-case scenario of the condition and was used as a benchmark for their own health as illustrated by these quotes:

I just always think of what happened to her, what she went through and I just think it's nothing compared to – you've got to take a few tablets. I don't worry and think. At least it's sorted. (Jane, F5, DCM, pos screen)

I'm lucky that I'm 41. My dad was 49 when he died and was on his third of fourth pacemaker…So I'm doing pretty well. (Maria, DCM, neg screen)

The prospect of dying early, based on their family history, was acknowledged by most participants. However, these two seemingly contrasting quotes demonstrated that participants work hard to cope with and process this possibility, again, heralding the need for better communication and supportive strategies around conveying sudden cardiac death risk in ICCs.

Considering obviously my dad passed away from the condition. I know he probably had a lot more going on, age and other issues related, slightly different, but yes, frightening really to think, "Oh hang on," then they start looking at you and thinking you now have this inherited condition. And knowing that somebody died of it at such a young age and not enough is known about it and you don't know what you're going to be like in five, ten years' time. (Pam, F3, ARVC, pos screen & PGT)

I know that one day it's quite likely to be fatal. I've come to terms with that on some level. (James, BrS, dec screening)

Whilst the sudden death of the proband caused profound grief, there was almost always the hope of something constructive arising from this situation. There was evidence of an increased sense of personal strength and resilience, and a greater sense of empathy and a desire to help others which forms part of the reconstruction and growth that follows bereavement (Calhoun and Tedeschi, 2001).

Just positive that something was going to be done. Although perhaps it was too late for my daughter, I wish I'd known sooner. But looking forward, you want to do everything that you can to save the rest of the family from going through anything that they needn't go through. (Linda, F4, DCM, pos screen)

When my dad died, 27 years ago, they didn't know. He just had a heart attack outside the shop. Okay, now you know that he died from that (Brugada), and you can help more people. So the more studies they do on us, the more they can help people. (Lisa, BrS, pos screen & PGT)

These accounts demonstrated how a sudden death due to an ICC in a family creates an imprint on individual and family life and was an important turning point in relation to the direction of family dynamics. No one was untouched by the death of their close relative and whilst they may deal with their grief individually, there was a lasting sense of bereavement and questioning of what could have been done differently.

In general, the sudden death brought the values of the family into sharp focus, bringing them together in support of each other to deal with the grief but also to ensure that all was done to prevent a recurrence. However, as well as strengthening family bonds, the death can also cause relationships to break down when there was a mismatch in a couple's depth of bereavement; or a retreat from social activities particularly if the deceased was an integral part of that person's social network.

By witnessing the worst outcomes of an ICC in the death of their relative, this sets the perspective of the family for what could possibly happen if they were diagnosed with the condition. This therefore shaped their attitudes to life, often harking back to how the deceased relative would have liked to live or how they would have liked them to live; and to not take things for granted. There was a sense of honouring the death of

the proband by adhering to clinical recommendations, giving it a purpose in maintaining their health. However, the feeling of risk for sudden death was ever present although some may appear to be calmer and pragmatic about it whilst others were more anxious.

Whilst families feel helpless in changing the situation for the proband, those interviewed seemed to move towards growth with most trying to derive something positive from the death of the proband, even going beyond helping just the immediate family and contributing to the wider health of others in their situation.

6.3.1.2 *Taking one for the team*-from disease comes prevention

In some of the participants' families, the proband did not die due to an ICC but was diagnosed following the survival of a cardiac arrest. Whilst the participant was concerned for the welfare of the proband, and initially focused on their subsequent risk for developing an ICC, there emerged a realisation that whether they got diagnosed or not, they had the privilege of being spared from the worst outcomes of the disease due to earlier implementation of preventative management in them and in future generations.

> *I think we were sort of told very early on when they realised it was Long QT, and they said to her, "It's a genetic condition, so we will need to test all the members of the family." And I said to Hannah, "Well thanks for taking one for the team", because now we all know we can all be protected, we can all do something about it…And I just looked at it and thought, well, we're incredibly lucky that a, Hannah survived, and b, that we know, and we know how to protect ourselves and future generations. (Julie, F1, LQTS pos screen & PGT)*

The psychological need of relatedness as described in SDT (Ryan and Deci, 2017) is fulfilled in these beliefs as the ICC creates a deep connection between those affected and at-risk and this began with caring for the sick relative and progressed to the belief that the proband has protected them from disease .

It is notable that even prior to the ICC being diagnosed in the proband, observations based on family history (including the proband at times) would already trigger general disease prevention strategies as observed by Jess whose mother was diagnosed with an ICC in her 90s and Fred who has a family history of multiple sudden deaths:

> *I am very focused on keeping fit, having seen from my mother's physical deterioration, not that anything is guaranteed. (Jess, HCM, neg screen)*

> *I've always been aware, because my dad died when he was only 50, and we knew it was something to do with his heart. Being aware I might have had a heart problem – because I know heart conditions can be genetic as well. So, I've always tried to keep myself healthy and things like that. (Fred, F1, LQTS, pos screen & PGT)*

Apart from relief that the proband overcame the challenges of their diagnosis, there was also gratefulness from the family that the diagnosis paved the way for them to have preventative treatment. This not only applied to existing family members but for any future offspring. In addition, family history also provided a cue to take up healthy habits for general health promotion transforming what could be perceived as a burden into a benefit.

The motivation for these preventative health behaviours in genetic diseases do not happen as a result of the knowledge of risk per se (Hollands et al., 2016), rather they are thought to come about by beliefs that changing behaviour can reduce risks and

that the individual believes they can change (Marteau and Lerman, 2001). This is congruent with the concept of competence in SDT, in addition to the strong personal ties or relatedness also seem to be at play in these accounts (Ryan and Deci, 2017).

6.3.1.3 Towards balanced parenting in inherited cardiac conditions

Parents of probands have a wide array of concerns and worries for their affected child and in these interviews, tended to describe in detail how they responded to their child's diagnosis. Parents of probands experienced shock and disbelief when the diagnosis was made mainly because of the child's apparent fitness and young age.

> *I think when we first found out about Henry we were just completely shocked…because Henry was into all sports, and he was the last person I ever thought would have had (a health problem) (Mary, F7, HCM, neg screen & PGT)*

Although an ICC is a chronic, serious illness, parents felt relieved at getting to a diagnosis following a range of non-specific symptoms, as medical management for the ICC would now be possible. The availability of management options appeared to have helped patients cope with the emotional distress experienced by most parents who are confronted with a diagnosis of a chronic condition in their children (Garwick et al., 2002).

> *There were some local boys that actually died, and I think that was making us thing a little bit more then…About, "Wait a minute, Henry has been getting out of breath for a long, long time." And the chest pains…I think when we got the diagnosis as well I think in a way it was a relief that it had actually been picked up. We felt reassured then, once it was under the hospital system we felt a big relief, really. (Mary, F7, HCM, neg screen & PGT)*

A key dimension in healthy parenting in SDT is autonomy support, alongside providing structure and being involved. Illness in a child is an added pressure likely to be rooted

in worry which is a result of parents' own psychological needs not being met (Ryan et al., 2006). Parenting can shift from being autonomy supportive to controlling as seen in these scenarios where initially, faced with a potential lethal diagnosis in their child, the immediate response of parents was that of extreme cautiousness and vigilance.

> *You could drive yourself mad over it (the diagnosis), couldn't you? It's just that nightmare (Pam, F6, ARVC, pos screening & PGT)*

> *Poor old Rachel, I used to get her duvet, if it was still and just shake it a bit. (Pat, F6, ARVC, pos screening & PGT)*

> *At the beginning…every time he said he was going to do something we worried about the risk element in everything he was doing. (Steve, F7, HCM, neg screening & PGT)*

Parents also played a major role in monitoring and ensuring that their children adhered to medical recommendations, and this applied even to young adults. Parents reverted to being more directive as they felt that their children may fall back or retain a carefree attitude which could impact on their health. May's description of her conversation with her son was not dissimilar to what he would have with a doctor or nurse.

> *I said to him, "So how do you feel?" And he said, "Oh well sometimes I get lightheaded." I said, "Well what do you do when you get lightheaded?" and that sort of thing, and he goes, "I go and sit down for a few minutes and get up and do exactly what I was doing." So it's like – and I said to him, "Are you taking your tablets regularly?" "Yeah, yeah." "Well how regularly are you taking them?" "When I remember." Okay. (May, F5, HCM, neg screen)*

Mary qualified her reasons for continuing to be closely involved with her son's care and her husband, Steve, supported her and can see where their son may need reminding.

> *Being his mum, but I still want to protect and go with him. And I suppose I want to have a clear understanding of what's going on. It's strange really, but you know, he's old enough to do all of this on his own isn't he? But it's very hard. It's because he lives here. If he had his own home I probably wouldn't be... (Mary, F7, HCM, neg screen & PGT)*

> *He is very forgetful with his condition isn't he? You still need to be around to ask all the obvious questions like, "Have you got your passport?" (Steven, F7, HCM, neg screening & PGT)*

This was the start of the parents' struggle to strike a balance between keeping their affected child safe whilst supporting the wishes of their children who were at the time, teenagers, and young adults. Parents did not lose sight that their children were at the stage of establishing their identity and acknowledged the frustration they were feeling.

> *She felt she'd been known by her school for her sport…She felt that was her, that was who she was. So to stop, she felt that she had to find a new identity. And she felt really angry (Pat, F6, ARVC, pos screen & PGT)*

Parents and affected children engaged in regular negotiations about what was allowed and not allowed within the ICC management recommendations, but children would sometimes still push their limits. These are extrinsically imposed limits based on the ICC and it was left to parents to provide an autonomy-supportive structure within which these are adhered to (Koestner et al., 1984). To this end, parents tried their best to take the child or young adult's perspective and provide meaningful alternatives. These are common dilemmas documented in the literature focused on parents whose children are affected by ICCs (Bratt et al., 2010, Farnsworth et al., 2006, Hendriks et al., 2005b). Mary and Steve describe how they tried to reach a

middle ground with their son's sporting activities, however, this excerpt illustrated how difficult it was to achieve this.

> *…we started to say, "Perhaps you'd better not do the gym." And he said, "Well if I can't do the gym that is the thing that's keeping me going at the moment." He said, "You don't want me doing the skiing, you don't want me doing the scuba-diving, but I want to do something. I want to do the gym." So we said, "Well, yes okay then, go to the gym. But don't overdo it." (Mary, F7, HCM, neg screen & PGT)*

> *But he overdid it. (Steven, F7, HCM, neg screening & PGT)*

Paul and Pat have tried to support their daughter, Rachel, to maintain activities safely with her peers as they know this was important to life as a teenager but accepted that there would be risks and that they needed to trust her to be self-aware. In the long run, this trust is likely to support Rachel in internalising values and regulation of behaviours that were not inherently intrinsically motivated (Ryan and Deci, 2017).

> *Yes, theme parks, we don't really encourage that. We've had to let her go once. (Pat, F6, ARVC, pos screen & PGT)*

> *She's 16 and I can't say, "No you can't." You just say, "Please don't go on the horrendous things that are really going to kick in the adrenaline." Alcohol, that's a worry. (Pat, F6, ARVC, pos screen & PGT)*

Apart from spending a lot of time dealing with the proband's medical care, parents felt that they should not get preferential treatment over their other children (Paul and Pat). However, sometimes parents felt so sorry for their sick child that preferential treatment was given as a way of making up for the child's sense of loss.

> *We gave Henry far more, and that's a bit naughty really, but it was the heart, it was actually that. We couldn't help ourselves, could we? We went, "Oh poor thing, he's got that as well. Let's spoil him." (Mary, F7, HCM, neg screen & PGT)*

The change in family dynamics was most often described when a sudden cardiac death occurred but this was demonstrated here where a severe form of an ICC was diagnosed in a young individual. It was evident that the consistent parenting structure that Mary and Steve have constructed for their children suddenly changed for the sick child. Mary described how the other children reacted to this scenario but as their son's condition became more stable, they were able to reflect on the effect it was having on the others and this practice stopped.

> *"You didn't do that for us. Why are you doing that for Henry?" "Well, Henry's got this heart problem and you know, it's not a nice thing to have is it?" Yes, we did use that as an excuse…we don't do that now, that was initially. (Mary, F7, HCM, neg screen & PGT)*

As the ICC symptoms became more stable in their children, both parents and children appeared to be more relaxed and less worried. They can live 'as normal' (Paul). However, this was only a transient calm, enough for parents to not think about their children's illness daily. As each follow up appointment approached, a sense of dread arose again and there was heightened anxiety and fear of how they and their children would react to any negative developments. Therefore, there was always a background level of anxiety and uncertainty which never truly disappeared (Gonzales, 2009).

> *When they said about the device, she got upset. And she said to me walking back, "Do you know? I just feel normal now. Enough time has passed, I'm starting to feel normal again, and then something like this happens." She seems to carry on as most of*

her friends do, to be honest. So we do feel like she has quite a normal life now. And I don't want to go to hospital and them say something that's going to change that and we're back to square one. (Pat, F6, ARVC, pos screen & PGT)

Long term, parents lived with the uncertainty of the health of their child but remained hopeful that medical advances would take place so that their health remained stable.

The only thing we always say is, "New things will be out in the future, they're inventing new things all the time." (Mary, F7, HCM, neg screen & PGT)

Despite the challenges and struggles parents had with their affected children, there was a huge admiration for the resilience of their children and gratefulness for a positive outcome despite their issues.

It was probably good for him to do what he did at university… because he had to be 100% focused on that for his study. So that helped, I would have thought. (Steven, F7, HCM, neg screening & PGT)

She turned it around. I kept saying to her, "Rachel, I know you've had to give up your sport and your triathlon, but you've got so much talent in other areas and you can go and explore that now." Thank God she has. She's done her GCSEs and she's given it her all. (Pat, F6, ARVC, pos screen & PGT)

Whilst parents spoke of their children overcoming their condition, they were very modest in their role in supporting intrinsic motivation through their provision of autonomy-support; and balanced structure and involvement that helped them achieve these successes (Ryan and Deci, 2017).

Parents ultimately tried to equip their children to enable them to manage their condition. However, this was difficult for parents whose affected child also had special needs. Shelia was in her 80s and remained the main carer for Peter who has HCM and learning difficulties. Whilst this role was becoming a struggle due her other co-morbidities, she spoke of the deep bond between them and the help they gave each other.

> *When we were on that television thing, at the interview at the end, they said, "How do you cope?" I said, "Well, Peter can't live without me and I can't live without him. We need each other." It's true. (Sheila, F2, HCM, neg screen)*

It is evident in these transcripts that parental instinct became even more dominant when looking after the proband with an ICC irrespective of the child's age. The tendency to be overly cautious and protective of their children is pronounced at the time of diagnosis which can be a source of conflict when the proband was also at a stage when they are seeking personal growth and independence. Once symptoms settled and there was a greater understanding of the proband's condition and management plan, negotiating and guiding interests to more ICC-friendly endeavours become major tasks for parents. These timepoints, as well as the proband's ICC clinic follow ups were periods when parental anxiety and worry were the greatest and when they would be least likely to consider their own health concerns as an at-risk family member.

In summary, this theme has revealed how the proband's story deeply affected every aspect of family life. This suggests that the knowledge and beliefs surrounding the diagnosis and in particular the sudden death of a proband, are held collectively within the family; and influences the actions and attitudes of at-risk family members regarding cardiac screening/PGT. This theme also brought to light the burden of guilt

and worry; and challenges parents must overcome to allow them to focus on their health and the wider issues in the family.

6.3.2 Theme 2: Leveraged autonomy

In general, family members at-risk for an ICC did not express regret at having made the decision to undergo screening/PGT. A presumption would be that the participants had made this decision after thorough weighing of the pros and cons of screening/PGT and consideration of how they would cope with results with the support of their healthcare provider. However, it was evident in the transcripts that many had come to the clinic with their minds made up on what they were going to do.

Whilst no one expressed that they were forced into their decision, which could be a concern for the younger adults still living with their parents, the reasons for proceeding were only reflected upon further down the line. Indeed, the transcripts from this study would be the first time some participants have elicited why they had undergone the process as summarised in this young family member's account:

> *You just went, "Oh, my brother's got a heart condition". You don't realise at the time that that's now affecting everyone else within the family, do you?...But then after that you think, "Hang on, there are loads of implications that come with it." Like, we've all got to be tested, and we have been. (Tim, F7, HCM, neg screen & PGT)*

The lack of regret to undergo screening/PGT gave a sense of autonomous decision-making amongst the participants but there were hints of coercion/overt encouragement by parents and a lack of a sense of choice in some participants. The difference between autonomous and controlled motivation is not dichotomous and there exists a continuum referring to degrees to which motivation is autonomous or controlled (Deci and Ryan, 2002).

In the following sub-themes, more detail as to how this autonomy was leveraged by motivations based on personal health, and family-based reasons, alongside the experiences of the process of screening, is discussed.

6.3.2.1 The pathway to screening and/or predictive genetic testing process

Participants described how knowledge of their risk for an ICC came about either from a health professional following a sudden death of a family member, after a suspicion of an ICC at post-mortem or information relayed by a relative or parent after receiving news of a diagnosis; and in one participant, second-hand from the carer of her elderly mother (Jess). For those whose only option was screening, this news started a series of events which included seeing a GP, getting referred and then being seen in a specialist ICC centre. In the case of those who had the option of PGT, they were seen by a genetic nurse or counsellor after a referral from the GP.

This process appeared to be a reflex reaction from most participants and when describing how they approached the screening/PGT process there was a sense that it was something that just happens as described by Pat:

> *I think it was just literally ticking the boxes to get where we are now. I didn't really feel much in the way of worried about it. I just felt that it's just something that has to be done. (Pat, F6, ARVC, pos screen & PGT)*

Paul and Pat have other children at risk for ARVC and as the case for most parents, the motivation for screening and PGT was less about their own health but to pave the way for screening & testing of other children and even grandchildren.

> *I don't care about me, to be honest. You just want to know that your kids are okay. That's it. If there's one thing you can hope for it's that they're okay. (Pat, F6, ARVC, pos screen & PGT)*

One mother (Karen) expressed that she would not have undergone PGT for LQTS if it were not for her children and this was the same attitude her sister took until she had children of her own.

The parental instinct that deemed screening or PGT as part of looking after their children is consistent across the ICC literature as reviewed in Chapter 3. This is no different in the participants interviewed in this study and with the application of SDT, we can identify this as an intrinsically motivated action with the full internalisation of the values they hold as good parents (Deci and Ryan, 2002). The challenge here is finding a way to also ensure that parents who are also at risk for ICCs take on board the implications for their own health and be intrinsically motivated to preserve it.

Even for those who did not have children, although personal health was cited as the main reason for screening, how their results contributed to the knowledge of ICCs in the family thereby helping in the care of other relatives was also a source of motivation.

> *But yes, I think it's a great thing. Preventative, I would have thought it would save the NHS money in the long run, if you can do something about something early on, I don't know…. I'm not going to have children, I'm 41. But my sister will, and my brother has already had a child. So if it is hereditary, we need to know. (Maria, DCM, neg screen)*

In probing deeper into the motivations for screening, participants spent more time in explaining why they sought a referral for cardiac screening/PGT. Some cited that an external but trusted influence 'set the ball rolling' for screening such as referral to a

specialist ICC centre (John). Jess needed a cardiologist opinion for her mother's heart findings before taking up screening as she felt her mother may not have relayed the correct information due to her infirmity and deferential attitudes towards doctors.

> *She has the approach I think, that a lot of her generation do, that inhibits her asking questions. "Doctors are at this level and I can't challenge them or ask them or whatever." Whereas I think, my generation and the younger generation tend to, because there's more information around, be more willing to question and find out more about stuff…I think because my mother's cardiologist recommended family screening, that I came. I would be silly not to, I just had to get on with it. (Jess, HCM, neg screen)*

Parents were a strong influence on taking up screening for both the young and old, but they have highly contrasting accounts of how this was initiated. The older adults, whilst they had other motivations for screening (their own children, personal health), mentioned in a light-hearted manner how their mothers were always prompting them.

> *My mother was badgering me as Peter had been diagnosed with it…I went along for the screening test, had the test done, mainly because she moaned at me, just to keep her quiet (Bob, HCM, unclear screen)*

For the younger adults screening was done as a family and they did not appear to express resistance to it.

> *I had no idea what was going to happen to be honest. It was fine, I'd go through it all obviously again just to be safe, but I thought it was fine…. It was never a big issue, it was just, "Well we need it done" and that's that. (Louise, F5, HCM, neg screen)*

> *I didn't actually think about it too much at the time. (Kate, F7, HCM, neg screen & PGT)*

Parents of older adults were able to be more explicit about how worried they were for their offspring and grandchildren and expressed their thoughts on the importance of screening and the possible adverse outcomes of not attending. In contrast, parents of younger adults needed to be calm and reassuring despite carrying a huge burden of worry for their children. These actions from both sets of parents could be misconstrued as controlling rather than autonomy-supportive but in the context of relatedness within families, this is rooted in caring and protecting their loved ones (Ryan and Deci, 2000). Furthermore, if it was the case that the internalisation of these values within the at-risk relatives brought forward the decision to have screening, then autonomy was levered by these factors rather than controlled.

For the young adults, Louisa and Kate's statements gave an impression of not having a full picture of the purpose and possible outcomes of screening. However, in the transcripts, most young adults acknowledged that they were aware that they attended the screening because of a family history of an ICC though the sense that they had a choice was not evident.

The sense of choice is emphasised in SDT as an integral part of autonomy supportive decision-making (Williams et al., 1999). As screening in ICCs can be a periodic, lifelong recommendation in the absence of PGT, this may lead to non-attendance once the young adults have left the family home.

For some participants, PGT became available after the proband's diagnostic genetic test results yielded a pathogenic variant for which they could be tested. Participants generally felt that this was a positive and definitive step which gave them more information about the ICC in the family.

> *It was that gene…I think maybe to begin with I wasn't really sure what it was and what it was that could be passed on. But yes, we then found out what it was and then we all could get tested for it. (Kate, F7, neg screen & PGT)*

However, some participants appeared to feel they had no choice but to proceed, with parental responsibility weighing in heavily, although it is implied that they could have declined in this account of the genetic counselling they received:

> *I just don't feel there was a choice in terms of the (cardiac genetic nurse) gave us the choice, from our point of view there wasn't a choice. To not take it would have been out of the question. (Pat, F6, ARVC, pos screen & PGT)*

The reflection of Mary and her children, Sharon, and Tim, around decision-making for PGT illustrated how important it was for parents to be autonomy-supportive, which included encouraging engagement with genetic counselling, despite desiring a particular action, in this case to take up PGT:

> *So I did sort of say to you all, didn't I, "I would really like you all to have it," …the opportunity is there to stop this happening to their children. But if any of them had said, "I definitely don't want to do it," then they wouldn't have had it done. Because they would make their own minds up with that. (Mary, F7, HCM, neg screen & PGT)*

> *For us it was never really an option not to have it done, because it if it's there, it's being offered and you know specifically what it is, then it gives you more of an insight and you can sort of, not prepare yourself, but you know more about it and you know to keep on top of it and things like that. It seems that for us it was a no brainer really, wasn't it? (Sharon, F7, HCM, neg screen & pos PGT)*

> *It's not like we walked in and it happened, we all sat down (and we discussed it with the genetic nurse) we sat there for a good half hour, 45 minutes, talking about it. So…nothing going against our will. (Tim, F7, HCM, neg screen & PGT)*

As a follow up to this, their sister, Kate, offered her personal view:

> *None of us sat there and said we didn't want to do it…But really – and I mean, as well, if (the cardiac genetic nurse) hadn't said about doing it I probably wouldn't have done it myself until later on, until maybe I was ready to have children. Then I maybe would have thought about more then. But it's good that (the cardiac genetic nurse) said about doing it when we could. (Kate, F7, HCM, neg screen & PGT)*

Whilst Sharon and Tim were more certain about their choice to proceed with PGT, Kate hinted at a possible desire to have had more time to think about it, although not regretting her decision. Again, it seemed relatedness and family solidarity played a part in leveraging Kate's decision-making. Whilst the others may have felt that they could make their decision around PGT within that 45-minute appointment with the cardiac genetic nurse, Kate could have probably benefitted from a period of reflection. This was evident as she expressed this opinion openly in the group session with her family but not when they attended the genetic counselling session together.

One participant decided not to have screening following the sudden death of his father from Brugada Syndrome. Whilst this event prompted him to come to the ICC clinic, he expounded on reasons for attending but not taking up screening at the time.

> *I guess just on balance it seemed as though if I were to have a positive diagnosis, the only thing that could realistically be offered to me that might make a difference was a defibrillator implant..and in particular given that I wasn't in a high-risk category because I*

wasn't showing any signs…it didn't seem like it would make much difference to me personally.

(My father) relayed very similar information to us about the condition and what the available treatments might be…I guess I hadn't had all the same information, but that same thought process had already been going in my mind for a couple of years. I think even when I came to visit I had it in my mind that I probably wasn't going to.

I think that was my main thinking in why I then didn't come to visit (the clinic) before his death then afterwards. It wasn't because I had had any kind of serious change of attitude, it was just so I didn't fall out of touch completely with (the clinic). (James, BrS, declined screening)

James' story was very detailed on how he took account of all the information that was available to him prior to coming to clinic at the time of his father's diagnosis. This was mainly from his father who he describes as a very learned man in whom he trusted. Whilst James eventually attended clinic after his father's death which was 2 years after being diagnosed, it was apparent that he was not going to change his mind about screening unless there were new management implications. At this time, he remained completely asymptomatic in which case, an ICD would not have been indicated. He acknowledged that the information from his father may have been different from what he could have gotten if he attended clinic earlier but his decision after his clinic visit is proof that, for him, his father's advice was consistent with standard clinical practice. This account demonstrated both an autonomy-supported environment provided by his father and his autonomous decision-making.

James remained well for a period of time but then started to get dizzy spells. He was aware of the signs and symptoms of Brugada Syndrome and promptly saw a specialist which was consistent with his decision-making as regards to his risk for an ICC-he

knew at this point a diagnosis could make a difference in his medical care. Concerns for personal health motivated him to seek medical care. The turn of events for James in the next excerpt illustrated how an individual may end up losing some of their autonomy as it becomes very difficult to control the cascade of events due to the clinical pathways in place to prevent sudden cardiac death in ICCs.

> *It turned out to be labyrinthitis, so completely unrelated. But again, as I said, I was basically getting very acute dizziness and sickness and raised temperature with that as well.*
>
> *At first the cardiologist that saw me when I was in for observation gave me the all-clear. But then referred me to another cardiologist separately, and I hadn't been expecting to be tested, but actually when I visited the cardiologist, he was a specialist in arrhythmia conditions and genetic conditions like this, broadly. He said just from a normal ECG it showed up clearly enough for him to make a diagnosis (of Brugada Syndrome). So I didn't exactly ask for a diagnosis, but I don't hold it against him because I was ill at the time anyway with something. (James, BrS, dec screening)*

James recalibrated his perspective on how his choices were taken away from him by rationalising it within the framework of healthcare pathways. At this stage in James' life, he has a partner, and his health decision-making started to become more family-centric, with the participation of his partner, at the prospect of having children. He continued to ensure he had adequate information before making decisions and increased engagement with the health care team.

> *As well as the ECG diagnosis I've had a separate genetic diagnosis, which identified a relevant mutation (for Brugada Syndrome). So we did discuss at one point with somebody…the possibility of having, I can't remember what the acronym was now, but something like IVF but with a pre-implantation screening. But*

> *apparently it hasn't been licensed for Brugada specifically yet. They think there would be a good case for licensing it but that hasn't happened yet. And we're at the stage now where we feel like we've got all the information we need for that and we're just thinking about whether we want to take that step or not. (James, BrS, dec screening)*

James was the only participant recruited who did not go ahead with cardiac screening when he attended the ICC clinic but rather than interpreting this outcome as a failure of the clinic staff at preventing complications of an ICC in this individual, it should be considered as supportive to his autonomy and competence. Through this engagement, he was more equipped to take actions for his signs and symptoms later. His story also highlighted the importance of emphasising that attending an ICC clinic or genetic counselling session does not mean automatically having tests as many have already made their decision prior to seeing a health professional. Just as important as having tests is having engagement with clinicians so that there is an awareness and discussion around the reasons for not taking up or delaying screening or PGT and so that any concerns and misconceptions may be addressed promptly.

Participants had various motivations for engaging in screening/PGT and these were consistently family-centric even if some did not have children of their own. The degree of autonomous decision making was therefore mediated by how much these family values were internalised rather than remaining an external factor for motivation. This is particularly important in young adults as they are likely to undergo screening for longer if PGT is unavailable; and have to cope with being a genetic carrier for an ICC or have a chronic condition for longer if they are subsequently diagnosed.

6.3.2.2 Screening and/or predictive genetic testing happens-ready or not

There was a sense of urgency for most participants to have screening or PGT. Indeed, participants were impressed if the wait to be seen and feedback of results in the ICC clinic was short and expressed disappointment if this period of waiting was perceived to be too long. Waiting was associated with a lack of knowledge about one's health status and was a common source of worry as explained by Jane as she compared her brother's experience in another centre to hers.

> *I think his GP did his ECG and then he had to wait for the result of that, because she had to send it to somebody. Then he went for this echo thing, which is another couple of months later, and then they didn't give him the results there, he had to go back to his GP. So, he had to wait, because you can't get an appointment with your GP very quickly, so he had to wait for that. Then they said, "Oh, there might be something there, so now you have got to go and see a cardiologist." That's six to eight weeks. So compared to my experience, he has probably got more time to be thinking about it and worrying about it. (Jess, HCM, neg screen)*

If cardiac screening was made available sooner, the wait for PGT to be made available to family members was not felt to be long despite sometimes taking a year from the testing of the proband.

> *Overall, we think it's all been dealt with really quickly, and we're very pleased, you know, that it's all come to light (genetic result) this quick. It's not been a long, drawn-out process, we don't feel it has. (Mary, F7, HCM, neg screen & PGT)*

Most participants described cardiac screening as 'straightforward' and a 'series of tests' (John) and did not feel underprepared for the appointment but when asked to describe their experiences in more detail, some participants felt there was a need to

provide clearer information on what was going to happen during their screening appointment prior to them coming to the clinic. Comprehensive information is a building block for competence which is a basic psychological need in SDT and the lack of it may result in patients going through a test without fully informed consent (Deci and Ryan, 2002). The participants' experience revealed that these information needs were addressed but tend to be dealt with on the hoof when they attended their appointments.

Whilst there was an expectation that some testing would be done, some participants were not aware of the specifics and practicalities of screening tests or realised to ask until they were already in the clinic. Often, the relief of just having an appointment dominated over other issues, such as long waits, and emotions associated with this, as evident in Jess' reflections, and so patients may not be as proactive in asking for information beforehand.

> *It actually took a bit longer than I anticipated for the appointment. There wasn't really any time. I should have asked, but I was still coming to terms with it or something. I thought it would be about an hour or so, and then they phoned up to say can you turn up a bit earlier? Then there was a lot of waiting around, so I miscalculated how much time it would be. So that information could be quite useful, you are going to be there for three hours, or something (Jess, HCM, neg screen)*

Participants asked to be forewarned of any crucial information required from them at the clinic visit as these may take some preparation on their part and can help clinicians do their job.

> *If you are going to be looking into people's history to say perhaps let them know- "These are the questions are going to be asked, if*

you're not sure it's really worthwhile trying to find that information out before you come," because (I was) questioned about dad's side of the family. (Val, F1, LQTS, neg screen & PGT)

Yes, and it would have helped you more as well. (Fred, F1, LQTS, pos screen & PGT)

Apart from knowing what tests would be done, participants also wanted to know why specific tests were being done and some practical advice to make sure they perform optimally on these tests. Female participants also reiterated the need to ensure their modesty when some tests required bodily exposure.

I'd already had an ECG…but I didn't know about the echocardiogram or what in particular they were looking for…this fitness test was just sprung on us, and I was like, "Oh God I haven't put the nicest underwear on." …I should have perhaps worn a sports bra today. Because you can't see into that room but you can see out but I was on the treadmill and thinking, "Oh my God, what can people see?" But just kind of being prepared for that…You just feel a bit more comfortable in something like that. (Julie, F1, LQTS, pos screen & PGT)

Competence as a basic psychological need in SDT not only entails having adequate information but also the ability to have confidence and readiness in taking action based on this information (Ryan and Deci, 2017). The lack of privacy and dignity will be a major hindrance to promoting this confidence. These young female participants have experienced distress due to the lack of preparation and inadequate care for their comfort and modesty which could have prevented their full participation in cardiac screening, if not for their concern for their health.

Sound clinical practice dictates that if there is a family history of an ICC then referral for screening and/or PGT is recommended. The participants seem to be aware of this standard and most desired that screening happened sooner rather than later. Due to the norms of long waiting times in the NHS, with an 18-week pathway as a standard, there was a sense that participants focused more on 'having an appointment' rather than the appointment itself.

Many of those undergoing cardiac screening only realised at the time of the appointment that there could have been more information and practical advice that would have helped optimise the appointment. Many might have had an idea of what was to happen, but some were not completely ready and found themselves getting attached to a machine for a cardiac test, going ahead with it, enduring discomfort; and asking questions during or after the procedure. This is extremely relevant feedback for clinicians as an ICC clinic visit will often have multiple components of tests and consultations but will fail in its aim to be comprehensive and truly 'one-stop' if patients are not given full information on what to prepare and expect on the day. This was a gap identified when ICC services were being established and it is evident more work is required to achieve this desired outcome for improved communication (Burton et al., 2010). These views were not expressed by those who had PGT perhaps because these tended to have a simpler format of a longer appointment (45 minutes) with one clinician and involved only taking a sample of blood or saliva.

6.3.2.3 Through the generations- concerns and actions for other at-risk family members

A diagnosis of an ICC created a ripple effect in terms of screening/PGT in the family and based on the results for first degree relatives, the recommendations will spread across the generations, and this can impact on the young and old. In the same vein that probands usually have the responsibility of informing family members,

participants who were diagnosed through screening or were found to be carriers, took on the task of informing relevant relatives.

The diagnosis of an ICC carried with it the burden of guilt and responsibility which motivated parents to encourage cardiac screening of others.

> *But obviously worried about my children and grandchildren, that's the concern…. I guess there's a slight guilt feeling that you may have handed something on to them. I think it's fair to say there's something there, that you feel a sense of responsibility. (John, HCM, unclear screen)*

Like John, all the participants informed their relatives who may be at-risk for an ICC and reported that this was an easy task and most of their relatives had, in turn, pursued screening/PGT. This was helped by having good relationships and existing lines of communication which showed that relatedness played a role in this process.

> *We are quite a close family. If we lived in one of those Indonesian longhouses, it would be ideal for us…It was just saying to my daughters, "Look this is out there, you need to get tested." Then it filtered down to the children and at their level it was telling the children, "You are going for a few tests." They didn't know what it was, it was just like saying, "You have got to have your eyes checked" or going to the dentist, exactly like that. But my daughters knew what it was, and their view was it's better to know and understand what you can do. (John, HCM, unclear screen)*

Relatedness could also be the reason why the positive views around screening were collectively held by John's family. When talking about screening with children, there was no option for not going and it was compared to a routine check-up that the grandchildren were used to. This message appeared to be misleading the children but at the same time seemed to be an attempt to normalise the screening process to

avoid instilling fear.

In the next excerpt, Fred is motivated by empathy to communicate the ICC risk with family members and because all his blood relatives have so far been found to be affected/carriers of LQTS, he has developed a belief that everybody else at risk will have the same results. This prevalence of LQTS has normalised the condition in the family so much so that the process of PGT and management implications are no longer considered onerous.

> *It was basically a no-brainer, it really was. And Hannah nearly died and I felt if it happened to my sister and she hadn't told me – so it was so easy, and I had to tell as many of my family members that are genetically linked to me about what's happened…Yes, sent an email or contact messages, and wrote to them and just told to them what happened, that it is a genetic condition; you've probably got the condition, get yourself checked, because if you know you've got it you can be put on medication to help. (Fred, F1, LQTS, pos screen & PGT)*

This perception of ease of access and management was widely held in the family as Fred's daughter, Julie, described the pace of screening for the children following the availability of PGT for LQTS:

> *She's (daughter) been seen and having the genetic test done within hours of her being born, and that process was quite quick…And just waiting for the results (of the PGT), and we've (her daughter) been seen already and she's already on medication. So I think now that we know, you know, for future family members, as soon as we have a new one arrive in our family we can kind of set the ball rolling. (Julie, F1, LQTS, pos screen & PGT)*

The belief of the family of their competence for dealing with LQTS may have been the basis for Julie opting to get pregnant naturally despite being offered PGD, which can be a lengthy and complicated process. These common beliefs can become a source of conflict if it is not held by everyone. Indeed, this became a major issue in Fred and Val's family when their ex-daughter in law did not hold the same opinion about PGT for their grandchild. The need for autonomy of their ex-daughter in-law as a parent impacted on this relationship.

> *But because his child's ECG is normal, by having that genetic test, it would put all of our minds at rest, and it's something that is very hard. My son has notified the school of the condition, whether the school have put a care plan in place for his daughter, I'm not sure. The GP is aware, so in terms of that, he has tried to cover it, as much as he can, and to the detriment of their relationship that he had with his ex-wife…It's got better but it's something – that is the one thing, that if we could change...And it doesn't matter how many times we tell her it's just a simple blood test, you know, her attitude is, "I'll let my daughter decide for herself when she's old enough whether she wants the test or not." (Val, F1, LQTS, neg screen & PGT)*

Although PGT in children for LQTS can be offered, sometimes health professionals will advise against it depending on the family's situation.

> *…And then we were counselled against it (PGT for the children), and one of the arguments was their autonomy. The little babies in my care, I'm overriding their autonomy. I actually still feel that…I completely understand the need for it in something like Huntingdon's or wherever where the mother and father are never going to be responsible for the care of the child; that's their decision, whether they want to look at their life through that decision or not….But actually I felt and still feel if you've got little*

creatures that you're in charge of, the more information you have about how to take care of them, the better.

But we ended up leaving it a little while anyway, mostly because I think at that stage we were in (the paediatric cardiologist's) care and he pointed out, and probably (the cardiac genetic nurse) as well, that they weren't in a risk stage. So that was perhaps our decision, to park it for a while. And then the decision about when it would be time to test them and talk to them about it. I found that quite odd, talking to them about it. (Karen, LQTS, pos PGT)

This initial genetic counselling conversation for the PGT of Karen's children happened at least 5 years prior to this interview and it remained a striking memory for her. Although the clinicians and Karen eventually agreed on a middle ground in terms of the screening and PGT of the children, there was a real risk of this relationship breaking down. Karen felt that it was worth looking back at the situation as it could have been handled better. For Karen, the session was clearly not autonomy-supportive of her decision-making as a parent although it is likely that the clinicians were advocating for the autonomy of the children. When Karen was challenged by the clinicians, this did seem to help her reflect and plan, and she was reassured that the children would be under regular screening. Indeed, she acknowledged that talking to her children about PGT was 'quite odd' however, this strengthened the supportive structure the children had as they embarked on PGT.

Frustration was not only felt when the younger generation did not take up screening/PGT, this was also felt when an elderly parent declined screening. In contrast to when young people declined, autonomy for this decision making was respected and accepted.

> *She keeps coming up with excuses, "I'm not having tests," …So when we said, "Have a test in London." "Oh, I can't, it's too far," which is fair enough. It doesn't give the whole picture though, does it, if everyone isn't tested?...Yes of course, it's her decision. I remember the first few times when I tried to get her to go for the tests, yes, I was annoyed, but again another thing I've come to terms with, really. I can't force her. (Ben, ARVC, unclear screen)*

Beyond immediate family, it was much more difficult to relay ICC risk information, but participants felt they had a duty to inform their relatives and made an extra effort. Jess does not have children, but she adopted the same instinct as the parents in this study and was aware that her adult relatives would want screening for their children.

> *We've just lost touch..They have children as well, I ought to try and track them down just to say about this. But also, between my brother and I, he's communicated with his son about it. (Jess, HCM, neg screen)*

Whilst participants who were parents either relayed the screening/PGT recommendations to their adult children or organised these for their younger children, participants who were found to be genetic carriers considered Preimplantation genetic diagnosis (PGD) for a future offspring. Preimplantation genetic diagnosis uses in vitro fertilisation techniques to create embryos which are analysed for well-defined pathogenic variants; only those free of the defects are implanted into the womb.

The genetic test bringing forward the possibility of PGD was considered a positive outcome of PGT and the three participants (Pam, James, Lisa) who were thinking about a family engaged in genetic counselling to find out more. This indicated that parental concern regarding a recurrence of the ICC also applied to future offspring. Only one of the participants had experience of PGD and this was for ARVC and

although this was not successful, the efforts of the team and the support of the genetic counsellor made this a positive experience for her.

> *We chose to go down that route (PGD) to give the child a chance of a normal life and not have to worry about an extra thing. Obviously that didn't quite work for me because you only have a certain amount of chances, but it's still amazing how many people are involved, how much research is done, how they can just make a particular test just for you…to get you there….The GC was amazing. The times I called her up, there were a couple of times I couldn't have coped…at one point you have an egg but you're still not allowed to have it because it has the genes and I desperately wanted that egg. I was just like, "Just give me the egg; I don't care." (Pam, F3, ARVC, pos screen & PGT)*

Pam's brother, Jo, had seen her going through the ups and downs of PGD and felt this was not something he would prefer for him and his wife, and considered an example of screening to manage a future offspring's ICC risk.

> *No, I just wouldn't want to go through the disappointment that my sister went through of it not working over and over again. I don't know how she's survived it. It would have been proper heart breaking… It's only like a 50% chance of the child having it as well. One of my customers…because her husband died of cardiomyopathy. She goes every year to get her kid checked and nothing has ever happened to him yet. (Jo, F3, ARVC, pos screen & PGT)*

Whilst failed cycles of PGD were disappointing, the prospect of terminating a carrier foetus was inconceivable for Lisa who was seen by a GC when she was trying to become pregnant. At the time, prenatal diagnosis for BrS, where the foetus can be tested for the pathogenic variant, was still offered for BrS. Currently, this has been put

on hold for most UK centres for BrS. Whilst it is uncertain whether PGD was discussed with Lisa, what remained in her mind was the prenatal diagnosis option.

> *..when I was getting pregnant they basically wanted to inject me through the stomach to see if the baby had the gene, but it was a high risk of losing the baby. So it was like, "I'm not going to do that, I'm trying for this baby. I tried for five years to have Matthew and he's finally here. You're not going to do that. What will be will be." (Lisa, BS, pos screen & PGT)*

Although, PGD or prenatal diagnosis is a way to prevent recurrence in an offspring, the participants in this study had an individual approach when these were offered and balanced their decisions against the possible advantages for the future offspring, the emotional turmoil the process would bring and their capacity to be able to look after an affected child. However, there remained an optimism that a child conceived naturally could be a non-carrier offspring.

The cascading of screening/PGT in the family was considered a logical and automatic step for many of the participants for their family. As a result of their experience, there was a normalisation of the process as a routine and easy pathway. Therefore, it became harder to accept when family members did not pursue it and there was a risk of not considering the autonomous decision making of their relatives. When beliefs are consistent amongst the family, this provides relief and harmony, however, when there is a difference in opinion and the family member declines screening/PGT, it can be a source of worry and frustration. Clinicians who advise delaying PGT in the children's best interest need to work closely and sensitively with parents as this can cause a breakdown in their autonomy in their parental responsibility.

In this theme, we can observe how the basic psychological need for autonomy was leveraged when patients make decisions for themselves and their families in the context of engaging with screening/PGT for ICCs. Family values, particularly concern for children, were internalised and was often the basis of the decision-making. Family concerns arose from the sense of relatedness that developed through the ties that bind families and this extended to more distant relatives with heightened empathy.

Whilst the participants were aware that they had a choice, many felt that their default action should be to go ahead with the screening/PGT for their family. Thus, autonomy was not thwarted in those who underwent screening/PGT-it was leveraged by the concerns for the family. It is, therefore, important for health care providers to provide support to help patients to also focus on their own health because there will be many more healthcare decisions along the line.

Many minds were already made up long before participants attended the ICC clinic and it seemed that there was a perception that one attends the clinic only if they were going to have a test. This notion risks alienating those who choose not to have screening/PGT. It is important to shift the perceived purpose of the clinic towards engagement rather than just a service to provide tests so patients are equipped with the right knowledge in case their situation should change. Therefore, it is crucial that relatives have comprehensive information on the purpose of the ICC clinic, the options, procedures; and any actions required from the patients, prior to their clinic visit to build their competence and confidence to optimise their attendance.

6.3.3 Theme 3: Harnessing competence

Competence as a basic psychological need in SDT refers to the need where individuals feel capable and progress to a sense of mastery over one's behaviour. People experience competence when they have a chance to be actively engaged and

immersed in their behaviour and feeling adequately challenged to grow and extend their skills and abilities (La Guardia, 2017). Perceived competence was already evident in some participants when they held the belief that ‘knowledge is power’ in taking the pathway to screening/PGT (Ben, Julie). Harnessing competence is not a passive process and must carry on beyond the initial period of testing. The following sub-themes illustrated the many ways this was supported and developed as well as thwarted and hindered, as the participants tried to cope with the results of their screening/PGT.

6.3.3.1 Baseline information, building competence

In general, the participants in this study sought multiple sources of information about ICCs. Some had done background reading prior to coming to the clinic and/or relied on their relatives for information but many sought out more detailed information after they received their screening/PGT results or were faced with decisions in terms of their clinical management. This demonstrated the innate desire for competence to help participants deal with the task at hand when it came to their health (Deci and Ryan, 2002) and knowledge about the condition would be the foundation.

Leaflets from the clinic usually provided by the BHF and patient support groups like SADS UK, Cardiomyopathy UK and CRY were very much welcomed by the participants and were found useful in giving background information on ICCs.

The participants obtained their baseline information about ICCs mostly from the internet despite most health professionals warning them against it. Some websites were felt to be more reliable than others such as the NHS website and the BHF. Participants had various views and reactions to the information available online but were generally alarmed by the focus on sudden cardiac death in ICCs.

Karen described the internet as a 'Pandora's Box' and that most people are compelled to find things through this medium. Which is why it is understandable that Lisa, like some of the participants, felt that they needed someone to help filter the information from the internet for them.

> *I don't think, sometimes it's not good, is it? My friend Steph said, "You're not going to go on Google, you're not. I'm going to go on Google. I'm going to read about it and then I'm going to tell you what it is". If it wasn't for her I'd have probably been straight on Google, freaking myself right out (Lisa, BrS, pos screen & PGT)*

For other participants, they tried to make sense of what these worst-case scenarios mean for them.

> *I did Google what hypertrophic cardiomyopathy is…There is quite a lot of information, it is quite reasonably clear. I think there is always a concern that basically they have a story about it being undiagnosed, and particularly younger athletic people, just basically keeling over…My impression was that if you hadn't keeled over when you were younger, you probably won't. It seemed to say it's less likely to happen if you got through to a certain age. (Jess, HCM, neg screen)*

Jess was processing the information by using it as a benchmark for her own situation. Older participants were relieved they were of a certain age and unlikely to suffer a young sudden cardiac death. Technically, this is correct due to 'young' as a qualifier but a sudden death from an ICC is rare but still possible in an older person.

May believed that a resource should be available from the clinical team to help mediate this information for patients and to explain what is applicable:

> *…they say don't they, if you've got something wrong with you, don't look on the internet. But that is the only place really you can find information about certain things. And also I mean you get so many different – it might be the NHS website or it might be somewhere else, or it might be in America, and they all have these different bits of information…So if you could actually speak to someone or send somebody an email and say, "I've read this, is it true?" And somebody can actually come back to you on an email or say, "Well look at this website, this will tell you everything you need to know." (May, F5, HCM, neg screen)*

The timing of information was just as important as the quality and quantity of health information available to patients. If they received bad news, they usually need a period to take this in before they can even process any information about clinical management. Some participants said that they were too distressed to take anything in during the early days of a diagnosis (Karen) and bad news makes everything 'foggy' (John). May described her own experience of arriving at a point where she could deal with the health information following a separate cardiac diagnosis:

> *… I mean you go through the "Why me?" And you go through the, all these people that have murdered people and they haven't got it…So you go through all these different bits and pieces…and I think until you get to the end of that process, and you've accepted it and then you want to know more about it, you're not going to take in any information. (May, F5, HCM, neg screen)*

May has made a near identical description of the stages of grief (Kübler-Ross et al., 1972) in her excerpt which emphasised the sense of loss that accompanied a diagnosis. Therefore, a single consultation is unlikely to address all the information needs of a patient following a result or diagnosis and a link to the clinical team is essential to help patients access this information later. In the ICC clinic, this link is

usually the cardiac genetic nurse (CGN) and although the CGN may be clear about their role and responsibilities, it may not always be obvious to patients, particularly in the early stages as Karen reflected on her experience:

> *I'm not entirely sure what the (CGN's) role is in all of it is, that maybe at some point things could be more clearly explained to people. But the only thing is, at the point of entry when I first met (the CGN) I was too distressed to properly absorb that kind of information, you know?...Who do you ring when something's not quite going right? Neither (my GP) nor his secretary knew and that makes a big difference…(Karen, LQTS, pos PGT)*

It was acknowledged that the internet has vast amounts of information and was consulted readily by the participants but with some trepidation. Websites and patient information leaflets were considered more credible if these were backed by the NHS or distributed by a health professional. Participants took on the general information from these resources but eventually started picking out which they felt applied to them or suggested that a health professional could help with this validation and tailoring to their situation. This initial information gathering provided the foundation for their knowledge competence around ICCs, but it was limited. In essence, whilst written and online sources were good for basic information on ICCs, the participants needed to draw on more resources to build up their knowledge and competence in dealing with the results of their screening/PGT or caring for an affected relative.

Timing was also an important element in taking in health information due to the emotional upheaval that often accompanies bad news. Apart from time and space needed to accept their results or diagnosis, patients needed to have a clear point of contact within the health care team when they were ready to access more information. Thus, in building perceived competence, adequate knowledge as well as confidence in accessing this knowledge, should be facilitated by the health care team.

6.3.3.2 Communication with health professionals-from the straightforward to mixed messages

Participants who underwent screening typically received feedback on their results on the same day as their tests. For those who had normal results, the consultations were short and straightforward which although efficient seemed more like a process than an interaction. This was not deemed inadequate as the results and plan were communicated clearly and it was felt that a more personal connection was reserved for people who had abnormal results.

> *…it was quite straightforward and said, "You've got no sign of this. Maybe get checked in five years' time" …. Basically, I was probably only five minutes with the consultant, because I guess if you have something he is going to spend a lot longer talking about this, that and the other…there would be more coming across as supportive or whatever. (Jess, HCM, neg screen)*

The health care team in ICCs is a hub of information for patients undergoing cardiac screening/PGT. Health professionals are relied on to give accurate and tailored information on two major aspects: diagnosis and management. In SDT, the patient-clinician relationship is a social context that should be autonomy supportive to ensure autonomous motivation and to increase perceived competence. This is supported by fostering relatedness between the patient and clinician through effective communication and considering the perspective of the patient (La Guardia, 2017).

A lack of relatedness in this professional interaction could be detrimental to maintaining regular follow ups for someone like Jess who must attend periodically for years. NHS clinics are inherently busy and overbooked so part of the role of the CGN

includes ensuring that whilst the interactions with the doctors may be brief, patients have a connection to the ICC team long-term.

> *I may not use (the service) all the time and I might not need to contact (the CGN); it's knowing that (the CGN) is there in case we do, because otherwise how do we get back into the system to get an appointment or be seen. I really do appreciate that really. That is good support. (Pam, F3, ARVC, pos screen & PGT)*

Communication of normal results or clearly abnormal results appeared to be an easier task for clinicians as participants moved on quickly to describing how they adjusted to their status. For those participants whose screening results were borderline, a few described the frustrations of receiving mixed messages, particularly from the cardiologists. Ben, who had unclear results for ARVC, described how opinions changed about his diagnosis with every clinic visit:

> *…what really stands out, so I had an MRI and a few weeks later I got a letter…it said, "It looks like, from the MRI, there's signs that you have got it (ARVC)." And so there I'm reading it on my own; it just felt a bit cold and impersonal…it all became very real at that point.*
>
> *And then I saw another doctor and they were like, "Ah, they shouldn't really have said that…With (an enlarged heart) alone, with no other signs, it's not enough to quantify you having it." And I was like, "Oh." So again, it was another weight lifted, but it's frustration because, "Oh, I've just been told," you know?*
>
> *I think I saw three or even four different doctors, I'm sure, and each time it seemed to sway from one to the other. The latest one, he kept saying probably not. But again, it's such a hard thing to diagnose, without other signs, without any really bad palpitations or anything. (Ben, ARVC, unclear screen)*

Ben also spoke about his frustrations on not being able to have a diagnostic genetic test for a possible ARVC diagnosis as his clinical picture did not quite satisfy the criteria for testing.

> *It felt like every family I'm sure would want to ask there and then but it doesn't work like that, again, I know now. At the time I'm thinking, "Why can't we just find out? Why can't we get the genetics test done?" (Ben, ARVC, unclear screen)*

ICCs are a relatively new disease entity and diagnostic guidelines continue to be updated as new discoveries for genotype and phenotype correlations come forth. New information kept changing Ben's likelihood of having ARVC and this made his emotions swing from one end to another about a pending diagnosis. This was made worse by contact with different doctors. The degree of relatedness between Ben and the doctors would make it difficult to foster an autonomy supportive environment but other members of the team can play a role in this. As time moved on, it is evident that Ben increased his perceived competence and insight on the complexities of arriving at a diagnosis. Ben cites the CGN as helpful in helping him process the medical information being relayed to him in clinic:

> *Going to see the doctor, and that was quite stern and then after seeing the CGN and that was a bit softer. It was nice to have a balanced time there. If it was just seeing the doctor and saying, "Right, let's see if you've got it, see you in six months." And it was all a bit harsh, but yes, the CGN made it a bit more comfortable... (she) told me exactly what to expect and when, and there was a lot of communication, I remember, that was good. (Ben, ARVC, unclear screen)*

After many years of screening, patients may be cleared of a borderline diagnosis due to updated diagnostic guidance or establishing other causes for the observed features

such as underlying hypertension causing myocardial thickening. After being managed as a borderline case for many years, Bob was told by his cardiologist that he did not have HCM. However, his beliefs on his current health status showed he needed more clarification on this.

> *Nobody actually sat me down and said what cardiomyopathy does. Yes, I read a bit about it, but you still don't know the severity of it. In my opinion I don't, but I won't accept it. To this day, as far as I'm aware, I have got it, but it has never ever materialised further. So in my mind I haven't got it. Whether it does materialise, I don't know. (Bob, F2, HCM, unclear screen)*

When an ICC diagnosis is ruled out, communicating this outcome must be done with great care as this usually comes after years of the patient believing they are going to develop an ICC. The patient has built a foundation for comprehension for subsequent messages based on this initial impression (Ley, 1988). Conflicting information, even if it is good news, may then contribute to greater anxiety, guilt, and misinformed self-images and decision-making (La Pean and Farrell, 2005). Bob related how this situation got worse when he subsequently suffers from a heart attack:

> *Then I had my heart attack, and as I say it is life changing. It's not just what you said, physically/mentally, it's the whole lot. It's just completely different. It's devastating… It makes you realise there are different departments and different aspects…I always thought, "You're the expert. It's your line of work. I am going to listen to exactly what you say." But, realistically, with my heart attack issues, I've grown to believe that's not actually the case.*
>
> *I don't look at doctors now, I still have respect for them, but I don't look at them as they are God. They are exactly the same as me; it's just that they have studied a little bit. As I say, the guy that I went to see about my heart issues, when he spoke to me in his*

office, I could have actually slapped him. He had sent me a letter saying one thing, and he told me in the office there was nothing to worry about. There is nothing wrong with me. (Bob, F2, HCM, unclear screen)

The main role of the specialist ICC clinic is to diagnose or rule out an ICC diagnosis, however, in the process, they are likely to see patients who have coronary heart disease (CHD) which is far more common than ICCs. It is normal practice to rule out CHD as a differential diagnosis, however, with the heart attack coming as a complete surprise to Bob, either this was not done or was done but not communicated well. If relatedness between the health care team and the patient is not well-established or disintegrates as in the case for Bob, it becomes even more difficult to provide an autonomy-supportive environment within which healthcare decisions are made and for Bob, the CHD brings forth other long-term health implications.

Patients need to have clear and comprehensive information, and this includes receiving bad news alongside good news to enable them to cope and make decisions about their health. In a relatively young field such as ICCs, the participants accepted that new developments happen all the time and may impact on the interpretation of their test results.

Good communication between health care providers and patients is integral to fostering relatedness which provides an autonomy-supportive environment for patients. This, and a full picture of their situation, helps patients develop perceived competence to cope with their risk or diagnosis of an ICC. Ideally, this relatedness should be established with the whole ICC health care team but due to constraints in healthcare provision-busy clinics, lack of time-this may only reach a superficial level with some, particularly doctors. However, as long as there is one member of the team,

like the CGN, in whom the patient can forge trusting and consistent relationship, this appeared to contribute to the sense of commitment of the whole ICC service and thus still maintained relatedness.

6.3.3.3 Participants who had a normal screen or non-carrier predictive genetic test results-going back to normal

A normal (negative) cardiac screen or a non-carrier (negative) result for PGT determines the next steps in a patient's ICC care pathway. A non-carrier result means that a patient is unlikely to develop the ICC and their offspring is not at risk for the condition. Participants, therefore, saw this a definitive test, releasing them from long-term screening and gave them relief (Kate and Tim).

In addition, those with non-carrier results felt that the experience helped them focus on their health and maintain healthy habits as a family:

> *Our own results? I don't – I suppose I haven't relaxed on that. As long as I'm okay then I try to stay okay or a bit better. I exercise more. I just like my biscuits. No, I think the junk food element has gone that we had. As Tim was saying, we do eat more salad stuff and walking. (Steve, F7, HCM, neg screen & PGT)*

A non-carrier result enabled Paul to go back to his exercise regime which he felt was essential to his well-being. This light-hearted exchange with his wife, Pat, hinted at how devastating it would have been for him if he was found to be a carrier. Pat's understanding of this and concern for him has somehow helped her accept her own carrier status that she can speak about this situation with humour, within a group:

> *I was probably a little bit relieved because of my sport…I did think I was going to have to stop (Paul, F6, ARVC, neg screen & PGT)*

That was really for selfish reasons. He likes his sport, so he can carry on…You were relieved…You could carry on as normal (Pat, F6, ARVC, pos screen & PGT)

The thing is, for me, I need my activities otherwise I'll go off my head. So it was kind of… (Paul, F6, ARVC, neg screen & PGT)

'Thank God it's you and not me' (laughter) (Pat, F6, ARVC, pos screen & PGT)

The main task for perceived competence for non-carriers was getting back to their normal life but also to try and enhance it with healthy habits. By the restoration of a sense of choice, volition, and freedom from the external pressure of the ICC risk to cut back on activities they enjoy (exercise), development and growth ensued. This autonomy supportive state helped individuals actively internalise the value of better health and thus were more motivated to take actions towards it (Ryan and Deci, 2000). To an extent, this was also exhibited by participants where only screening was available. Those who had a normal screen reported that they were happy and reassured by their results and had 'no excuses' for decreasing their exercise regime.

I shall be trying a bit harder in my fitness thing. Whereas before I probably maybe pulled back a bit because I didn't know. Afterwards I thought I can carry on now, as normal. (Jess, HCM, neg screen)

However, their relief is not complete as they were aware that there was still some risk that they would develop the condition in the future. They accepted the situation but made sure they knew what to do should their situation change, particularly for those who were younger:

> *It just makes you a bit more pragmatic about things. If it's going to happen, it's going to happen. There's nothing you can do to stop it…I don't think I'm thinking about it because why waste your time thinking about it? But yes, I know what to look out for. (Maria, DCM, neg screen)*

From the accounts of the patients with normal screens and non-carrier results, the relief that they mentioned after getting their results implied a sense of being coerced by the risk for an ICC as they stopped or they did less of the activities they enjoyed, particularly for Paul and Jess. This was a threat to their autonomy which was resolved by normal results (Deci and Ryan, 2002).

The period where patients receive normal results was an opportunity for personal growth and health promotion as the resolve to live a better, healthier life emerged. Therefore, patients would be more receptive to general cardiac health advice on diet, exercise, and smoking cessation at this time.

Whilst a normal cardiac screen/non-carrier PGT result brought relief, it was quite clear that those who could not have PGT yet knew that there was still the possibility of developing the condition. In these participants, to enable them to accept and cope with this potential cause of constant worry and anxiety, they acquired key knowledge and skills to develop competence to manage the uncertainty. This included the knowledge that screening was an ongoing process, and they developed the confidence to access it; and knowledge about what signs and symptoms to look out for that signal deterioration. This brought back the sense of control and autonomy that was being threatened by the ICC risk (Folkman et al., 1986).

6.3.3.4 Participants with a possible or definitive inherited cardiac condition diagnosis and/or genetic carriers-getting on with it in hope

The initial reaction of participants regarding their diagnosis, likely diagnosis, or genetic carrier status, was shock because in common with most patients who come for screening, they were well and asymptomatic and expected normal results. The initial shock was followed by processing of the information and trying to discern their status in the ICC disease continuum, mainly trying to figure out if they were at risk for sudden cardiac death. To do this, participants took stock of their personal health, and compared their status to textbook descriptions of sudden death in ICCs, the proband or the sickest member of their family and/or the oldest, least symptomatic family member who was in the same situation as them.

> *For me personally, it made no real great impact I don't think, emotionally. I've survived this far, so I have had pretty good life. (John, HCM, unclear screen)*

> *…my mum is a carrier. She's doing pretty well. She's very active…Had a few things, but that put my mind at ease to see that she's – and her parents were okay. My granddad, her dad, lived to 94. That all kind of bodes well from my point of view. (Pat, F6, ARVC, pos screen & PGT)*

This was consistent with the practice of risk stratification in the ICC clinic, which is the next step after a diagnosis or positive carrier result, confirming that ruling out the risk for sudden death is the top concern for both patients and clinicians. Caution must be advised for comparing against relatives as variable penetrance is rife in ICCs and these presumptions of having the same outcomes as relatives may not materialise which Karen had noted. Furthermore, it is important to note that there are other health implications that may impact patients profoundly and many adjustments may still be required of them.

The majority of the participants who were diagnosed with an ICC or possible ICC through screening and those found to be carriers through PGT were asymptomatic or had very few symptoms. In the management of ICCs, particularly in LQTS and ARVC, lifestyle advice includes some exercise modification even if the patient is well. In LQTS, beta-blockers are recommended as primary prevention for asymptomatic carriers. Most participants felt that these management recommendations were reasonable, worth doing considering the possible consequences and they were not perceived as coercive to their lifestyle (Jane, Linda).

Participants described what they did in terms of adjusting their lifestyle and this was done mostly by continuing to do what they enjoyed balanced with the extent to which they remained well or started having symptoms whilst doing these activities rather than based on medical advice *per se*. Some of the following excerpts hinted at a loss of autonomy due to medical recommendations or symptoms. However, we also see how individuals developed their competence in assessing their situation then gradually show willingness to act within these constraints in alignment with their values (Dworkin, 1988).

> *I used to run six miles like three or four times a week and then (the cardiologist) did actually say, "Cut down, you shouldn't be doing that much because that is pushing it a little bit." I did do my bike ride, which was 54 miles from London to Brighton and actually that wasn't a good idea because towards the end of that I was actually passing out…I really, really wanted to achieve it, but I raised money for the British Heart Foundation….I really did want to achieve at least half a marathon at a pace, but I've been advised that is not a good idea either. Maybe I will get back to a little bit of running but just not as I was before. (Pam, F6, ARVC, pos screen & PGT)*

I mean to be honest I haven't really changed a great deal of what I do. I'm a bit more aware if I actually go to the gym, which I haven't done for a little while, but just make sure that if I'm doing a class I tell the instructor…I don't have any symptoms or anything, so just make sure I take medication and that's it really. (Julie, F1, LQTS, pos screen & PGT)

Karen struggled a bit more with the need for medications as she felt *'big pharma was going to take over her life, for the rest of her life and make her take hardcore medicine'*. Her personal values were not aligned with what the medications represented. However, her personal circumstances had changed and alongside, a GP who supported her decision-making helped her see how the beta-blockers could help her.

...there was a GP who said to me, again this was after Jim left and I was quite stressed, but you know when you're really stressed but you don't know you are? This GP said, "Well look, you won't take sleeping tablets and you won't take anti-depressants. The only other thing I would suggest to you is Beta Blockers, and you have some of the best surgeons in the world telling you to take Beta Blockers and you won't take them." So I went, "Okay, I will." It made sense.

…then also because I knew that the girls tested positive and that was prescribed for them, that I was not going to choose no. And then I thought that I'd also like to know what I'm giving them, so that helped me make that decision. (Karen, LQTS, pos PGT)

If Karen was not having a stressful time, it could be argued that she may have never taken the beta-blockers but a further motivator for her to adhere to recommendations was her values as a mother which has consistently been the core of her decision-

making all throughout her experience with PGT. This concern for family may sometimes come across as coercive, particularly in a spouse/partner relationship:

> *Since the diagnosis, yes, apart from the worry for my family, I don't particularly worry about myself…I mean one of the things I used to do is scuba diving. Val says, "You can't do that now because you have a heart condition." My argument is, I've always had a heart condition because it's genetic. But, the voice of reason is, actually should I do it? No, I shouldn't. (Fred, F1, LQTS, pos screen & PGT)*

In Fred's case, he considered the medical advice as well as the opinions of his family in adjusting his physical activity. Although Fred appeared to be arguing for his right to go scuba diving there is no evidence of being forced to stop, as he came to the decision informed by his own sensibilities and values (Ryan and Deci, 2004) specifically, his conclusion that it was the right thing to do by weighing the possible consequences of scuba diving for himself, as implied by the term 'the voice of reason'.

None of the participants in this category were competitive athletes nor was sport described as part of their identity, hence, it was easier for them to accept a recommendation for a less intense form of exercise. However, it was evident that if exercise was a part of a patient's regular lifestyle regime pre-screening or PGT, most would want to continue with it in some form or manner.

> *Interviewer: So they've given you that advice (about exercise), have you thought about it and changed?*
>
> *Yes, I think about it. I'm determined to carry on. (Bill, F4, DCM, pos screen)*

Bill has only been recently diagnosed and was given initial exercise advice and at this point he perceived this as a restriction. To help support his competence to evaluate and adjust his exercise regime, an autonomy supportive approach should be taken by clinicians to preserve a sense of choice and avoid feelings of coercion.

Unlike Bill, many of the participants have known about their diagnosis, likely diagnosis, or carrier status for quite some time. During the interviews, they reflected on their past and current attitudes about their health status and how they coped with the challenges this brings.

Ben's excerpt encapsulates how a profound period of reflection helped him examine his strengths and weaknesses and give himself space to be vulnerable. Only then was he able to gain perspective on the situation at hand to be able deal with the uncertainty of his diagnosis and gain a positive outlook.

> *I felt like personally I had to dig deep to get through it. And that's stayed with me as well, it made me stronger. It's given me more understanding, yes. I'm not saying I'm a saint, but you know?*
>
> *Nobody likes the uncertainty of not knowing, nobody likes to be unsure. But yes, it seems to be a smaller problem than it was. It sort of hangs over you in one form or another in the back of your mind. Probably most days you think about it. But yes, it's pretty good at the moment. I'm trying to be positive about things.*
>
> *I've changed quite a lot as a person in the last few years and I think for the better. Yes, again a little soul searching and lifestyle changes, being healthier. (Ben, ARVC, unclear screen)*

The ability to do a reflective appraisal to a high degree like Ben can deepen the sense of self-determination and autonomy (Dworkin, 1988). Ben also mentioned self-help

books helped him facilitate this reflective practice but as time went on, he was able to do this spontaneously. As an individual evaluates and identifies with particular desires, goals, and opinions, they can endorse or reject them. Those that they endorse wholeheartedly become part of their identity and distinctive self which spurs intrinsically motivated actions to achieve these wants and goals (Friedman, 2003). In Ben's case, he has achieved a sense of calm despite the uncertainties about his diagnosis. He acknowledged that there are still difficulties but was able to take actions that promote health.

Similar to participants where only screening was available, those who have a diagnosis, unclear screening results or those who were carriers felt that the presence of the ICC and its ramifications were 'still in the background' (Pat, Ben) and on a practical level, they needed to have strategies to keep these thoughts from their mind on a daily basis. For some, like Lisa, this involved taking a course which provided a tangible output as well as distraction.

> *I thought, "I need to do something with my life." So I went and did a beauty course...two years at college training to do it. I came out with this qualification. It may be the best money I spent, because it took my mind and made me appreciate everything. You just need that little something to do, don't you? (Lisa, BrS, pos screen & PGT)*

In these statements, Lisa not only demonstrated how her actions helped prevent thoughts of her risk of sudden death to dominate her life, but also how competence is experientially significant to the self. By taking ownership of the activities by which she gained her achievement, her perceived competence nourished her sense of agency and ability to cope with the challenges in her life (Deci and Ryan, 1985).

Others took a more fatalistic view of possible events to come because ICCs are genetic and some of these events can be unpredictable. Focusing on the here and now helped them carry on day by day but from earlier excerpts there was some evidence that these participants have developed competence in evaluating signs and symptoms that may herald disease progression. This awareness combined with confidence in knowing when and how to seek help was key to coping similar to the group who had normal screening.

> *There are two main things that this heart condition, so far, has had on my life. One is that it might kill me one day, right? Without wanting to be too blunt. But it's not affecting me in the sense of me having any symptoms. But I know that one day it's quite likely to be fatal. I've come to terms with that on some level… sometimes I'll say, "I had 26 years of my life where I got by just fine without a diagnosis." I kind of look to it more like that, that I've got, hopefully, plenty more years ahead of me where it's basically had no impact on my life, and I try to not let the label itself affect me too much. (James, BrS, declined screening)*

> *Well, over the last few years...I tend to just forget about it and get on and live your life. Whatever is going to happen is going to happen and you don't know what's going to happen so just kind of get on with things really…what is the point in worrying about something that you don't know is going to happen? (Pam, F3, ARVC, pos screen & PGT)*

Notions of fatalism may counter beliefs in the ability to control factors that could minimise risk of mortality and morbidity in ICCs (Walter and Emery, 2005) but in these excerpts, a fatalistic attitude helped counter the daily worry of sudden death that was common in the participants. This sense of worry was recognised by participants as a real threat to their mental health and indeed may contribute to more symptoms. They resolved to overcome these thoughts or decrease the intensity in their lives. Many

spoke of an attitude of ‘getting on with it’, ‘dealing with it’ and carrying on with a positive outlook:

> *I say this with all people with handicapped kids; you deal with it, you forget about it. If you kept it in your mind all the time you’d go mad. That’s what happens. You get so used to things happening that you just cope with it and that’s it. (Sheila, F2, HCM, neg screen)*

> *I think you have to stay positive, don’t you? And keep looking for happiness in your life…but it’s always there in the background. You can’t get away from it, it’s always going to be there. I suppose it’s just learning to live with it in the best way that you can, and to get it to fit into your life in a way that you can manage it a little bit. Very occasionally I will just get into really silly thoughts, but it’s so, so rare now…definitely sometimes if I think about it I start to feel sick. That’s the worry. (Pat, F6, ARVC, pos screen & PGT)*

This show of grit and determination demonstrated a strong intrinsic motivation to preserve psychological well-being as this would have consequences personally and for their family life. This motivation may have come from an external source, such as the ICC, but it has been fully internalised in concordance to what they hold dear. Pat explained how this sense of calm can easily unravel therefore, individuals must have the competence to regain their perspective on the situation and have the knowledge to act accordingly with support from the ICC team. An important timepoint for participants was during their check-ups. Ben said he prepared himself for the worst news whilst Lisa explained this was when doubt and worry heightens and spirals again.

> *When I have my appointments, that’s when it starts to play on my mind and you think about things. But then a normal day, I don’t*

even know I've got this life. I'm just a normal person. I do have an issue with death. I think it's all because of this (the ICD). I don't know whether – I think I suffer a bit with anxiety, and then I start worrying about people around me. And I shouldn't be worrying. And then I start worrying about my partner and, "Why am I worrying for that?" (Lisa, BrS, pos screen & PGT)

Lisa was one of the two participants with a primary prevention ICD and neither of them have had any treatments/shocks thus far. For Lisa, who was in her 20s, this was a constant reminder of her risk for sudden death which triggers her worry for her young son and partner who she will leave behind. However, during the interview she contrasted on how different her situation was at the time of the interview and how she felt lucky to have a diagnosis. She had initial fears when she was told she needed an ICD implanted but subsequently felt that having an ICD scar was a positive symbol.

"I'm a young girl so I'm going to have a scar across my chest." That's what you think then, but now, like I said to you, this holiday, I can walk along with my bikini top on. I don't care, because you might have it (an ICC), but you don't know. I'm just lucky. And if that's what the scar's gonna cause and I'm not fazed… (Lisa, BrS, pos screen & PGT)

Follow up appointments were also a source of reassurance for participants, particularly when they received good news or there was no change in their situation. It was a positive reinforcement for all the adjustments they have been making which builds perceived competence and a feeling of safety in the knowledge that the clinical team is monitoring them, which strengthened relatedness.

Health wise? Yes, I feel quite reassured that both myself and my children are being monitored and that we're in good hands obviously. I've had sort of emotionally managed to park the scarings of what, of

the ticking time-bomb, and just get on with things. I don't know if that is time, or if it's a possible question of sanity because you can't look at that every day. (Karen, LQTS, pos PGT)

Karen acknowledged that there was a clinical team supporting her and her family which gave her reassurance for any eventuality. It was evident that this has helped her immensely and the relatedness between her and the team was strong as she spoke of somewhat defusing a 'ticking time bomb' which enabled a normal life. In common with other participants, Karen also cited that perhaps time was an important element to coping as patients acquired competence and experience with living with ICCs as time passed. Val and Julie recalled how they felt at the start of their ICC journey and how over time and gaining a wider perspective, they were able to live relatively normal lives:

It was, at first so, so stressful to live with, you know? And I can remember I felt the most devastated, not when all three of my children were diagnosed, it was my grandchildren. That was the hardest. That it's hard when you look at Hannah to realise that actually five years ago that did happen, and it's just a case of sort of having to take medication and we know that the family are protected. (Val, F1, LQTS, neg screen & PGT)

I think recently I was watching a documentary on television and there was a guy who had a cardiac arrest and I don't know, I think because we've all come to terms with it and living with it, I was so shocked at the stats… And I was just sitting there thinking, "Wow, I'd forgotten how absolutely lucky we are" (Julie, F1, LQTS, pos screen & PGT)

Participants who had a diagnosis, possible diagnosis or were found to be carriers for an ICC mostly remained physically well following their results but the psychological impact of the possibility of sudden death was the top of their concerns. Adjustments

to lifestyle recommendations such as exercise and medications were eventually adhered to, but this did take some time to accept especially as patients did not feel unwell. The decision to follow medical advice was made by weighing out the advantages and disadvantages of adherence, degree of impact on current lifestyle, and the presence of signs and symptoms.

The prospect of sudden death remained a backdrop in their lives, even after many years and in the context of very few symptoms. However, as time passed, it became less of a dominant feature day to day and participants employed various strategies to achieve this. The first step was to acknowledge that their psychological health was threatened by persistently thinking of sudden death, then participants employed distraction strategies, reflection, and the resolve to 'get on with' things. Eventually, as they gained confidence and competence in recognising danger signs, knowing when and how to access the clinical team, adjusted to the medical and lifestyle recommendations; and experiencing very few health issues, a sense of normality and hope prevailed; and some even considered themselves lucky despite the diagnosis.

6.3.3.5 Variable penetrance in inherited cardiac conditions requires tailored health advice

The unpredictability of outcomes once a person is diagnosed or found to be a genetic carrier for an ICC is well-known. None of the participants experienced a cardiac arrest after receiving their results but a few showed signs and symptoms, mainly palpitations, chest pain & breathlessness. Prior to coming to screening, they would not have normally taken notice of these but now with the ICC in the background, participants, tried their best to discern whether this was associated with the ICC or not as in this excerpt from Pam:

> *It's like if you've got a dysfunctional kidney, they can remove it and go, "Okay, you're okay now. You can recover and you can get on with your life," whereas this you just don't know, do you? It is one of those things and you will never properly know because like I said, each person is different. Sometimes I think, "Ooh, I'm getting pains and it is on that side, is it related? Is it not? Is it just muscular?" Then you think, "Am I being a hypochondriac, am I not?"*
>
> *I could see (my heart rate) used to go up to 3,500 beats and now all of a sudden it's 5,000 but they're (the doctors) saying, "Oh but that's okay." …that is a big jump and to me, things have changed but nobody has explained that to me properly and I did get worried. Does it mean it's got a little bit worse? I'm not saying it's got drastically worse but does that mean there has been a little bit of a change? Am I to expect that in another five years' time? Sometimes you don't want to feel like you're asking too much and too many questions. (Pam, F6, ARVC, pos screen & PGT)*

Pam recognised how unique each person's experience with an ICC is and whilst she was confident at recognising the signs and symptoms of an ICC such as chest pain, she was less confident about whether to seek medical care for this or not because she knew there could be other unrelated causes of chest pain. Pam was also finding it hard to gauge how much a change in heart rate is acceptable or not in her situation. In her mind, an increase from 3,500 to 5,000 beats per hour was a big change and a sign of deterioration, however, this still corresponds to a normal heart rate. As she developed her perceived competence for recognising symptoms for an ICC, this diminished as she became frustrated with her difficulty in distinguishing whether these symptoms were truly problematic. Pam then hesitated to approach a healthcare professional to get some reassurance as she did not want to bother them with what could be a trivial matter but could equally prove detrimental if these symptoms needed prompt attention. Therefore, to promote perceived competence, it is fundamental in a patient-clinician relationship to support the ability to recognise possible signs and

symptoms of an ICC as well as foster a sense of relatedness where patients will have the confidence to discuss these concerns without feeling that this is an imposition. This relatedness becomes even more poignant for those who do not have continuous monitoring devices implanted as periodic screening gives reassurance only for a specific period.

> *I can feel my heart playing up every now and again, and I don't know, is that Long QT or is that normal, or is that – and it's never been caught on tape or a monitor that I'm aware of. So I suspect something is at play, but I feel reassured that it's being monitored…And then the knowledge that it could quite easily be not being monitored, you know? Because if there is an issue, it's that thing isn't it? (Karen, LQTS, pos PGT)*

Karen was reassured when her periodic check-ups revealed that her symptoms have no associated signs 'caught on tape or a monitor', however she was very aware that most of the time she is not monitored so as in Pam's case, confidence to access the ICC team would be supportive of her perceived competence in dealing with her symptoms.

There were instances where it was not symptoms patients were worried about but changes in their bodies that may be a side effect of medical management or lifestyle recommendations. This also requires tailored advice from the ICC team as participants' intrinsic motivation towards health becomes compromised when the actions recommended for this has an undesirable effect.

> *It's just annoying because only time can tell…Like I can try changing that tablet, then I've still got to wait six months to see. Nobody can do anything about it, it's just the way that your body is. You can't speed it up. But I'm looking forward to going back*

> *and letting them know that my weight is still the same and then maybe swapping the other one and hoping that will change things. Because I just find it really hard to lose weight. (Jane, F4, DCM, pos screen)*

Another area in need more personalised advice is exercise. Whilst competitive and endurance sports are generally not recommended in ICCs (Pelliccia et al., 2020), the concept of 'moderate exercise' as a general guide for ICC patients is a vague notion, particularly as ICCs can affect people at any age with varying exercise capacities. These excerpts demonstrated the dilemma of trying to get levels of exercise right in light of an ICC diagnosis:

> *I think I have become a little bit more aware, if I am exerting myself and get out of puff, "Is that okay? Am I pushing too far?" I possibly would not exercise to that extreme that I would have before. I am a little bit confused as to whether it's just a feature of age, because you slow down as you get older. Because I look at people who are a similar age or a younger age and they seem to be in a worse state than I am, so I don't know what to judge against. (John, HCM, unclear screen)*

> *Obviously they've only discovered this condition, they don't know much about it. So I suppose they can't go to somebody, "Don't do this and do that." It's hard for the consultant as well, I understand. But I felt a bit like should I carry on doing my Pilates or not? What do I do? It's like, "Oh yes, do it, just don't do it too hard." I was like, "I'd just rather not do it, because what is too hard?" I don't know. (Jane, F4, DCM, pos screen)*

Whilst participants were aware that there might be limited information that could be offered to them in terms of exercise as ICCs are still a relatively new disease area, there is a risk that patients are either doing too much or being restricted to too little exercise without more input from health professionals as to the appropriate level of

exercise for them. Survey data suggests that patients with HCM in the United States are less active than the general population and patients purposely reduce the amount of exercise that they do (Reineck et al., 2013).

The ability to be physically active is widely accepted as an indicator of health and is also crucial for developing a sense of well-being, thus, it is not surprising that most participants mentioned how they fared with sport and exercise when asked about their health (Subas et al., 2018). Therefore, increasing knowledge in the quantity and quality of physical activity of ICC patients can undertake using a more tailored and practical approach offers a huge potential in promoting perceived competence and autonomous decision making within the broader guidelines for their ICC which has recently been updated and refined (Pelliccia et al., 2020). Indeed, many experts in the field are calling for a shared decision-making model when it comes to exercise advice (Baggish et al., 2017) and this should apply across patients with ICCs, not just athletes.

6.3.3.6 Accessing psychological support-It's a good idea for some

Most participants, whatever result of their cardiac screen or PGT, spoke of some psychological aspect of being an at-risk relative whether this was before, during or after their screening. Many spoke of the emotional upheaval their families have gone through with a sense of 'getting back to normal' often only after a few years after an ICC diagnosis via screening or carrier status result. Only one participant (May) accessed bereavement support after the sudden death of her son from an ICC, and her son (Tom) received some counselling via his school.

> *Once the funeral was over, it's like I didn't have anything anymore. And it was like, I kind of did fall apart then, which wasn't good. But my work had got in place this thing where you can go and talk to people…and they sent me to a bereavement counsellor which*

really, really did help…I was trying to be strong for them and not show them that I was upset, but with somebody else who didn't know me and what have you, I could basically blurt out whatever I wanted to do. Or just sit there for an hour and bawl my eyes out, which I couldn't do in front of these (my children).

I think these (my children) should have had some sort of counselling, but they didn't so, I think some of it is actually coming out now as well, I mean I've noticed with my other son, Ray, he's had stomach issues and I think he's getting a lot better, because he spoke to some counsellor. I mean obviously I don't know what he spoke to him about, but I think that was something to do with build-up of different bits and pieces.

You've (directed at Louise) had different things as well where you've gone off the rails a little bit, which I think goes back to not being able to talk about things.

(Addressed to Tom) You know after Mike died and we went for that meeting, if they had offered bereavement counselling, would you like to have gone? (May, F5, HCM, neg screen)

I don't know, probably not, I don't really talk about my feelings to be honest. So, no. (Tom, F5, HCM, neg screen)

May sought out the counselling herself as she knew this was being offered at work but not through the clinical services where the family was already undergoing screening. She talked about how helpful this was when all the activities around the autopsy and funeral had quieted down as this busyness was what was holding her together. Her counselling sessions were a neutral place where she could let go of her emotions, show her weaknesses, and did not feel judged whilst with her children, she had to always appear composed and strong to support them. She felt that some of the social issues that her children have gone through or experienced was a manifestation of not having had the opportunity to discuss their feelings with a

therapist at the time of Mike's death but doubted whether they would have taken it up at the time.

Post-traumatic stress in mothers whose child has had a sudden cardiac death due to an ICC is well-documented (Ingles et al., 2016) and May's own competence in recognising the need for psychological support prevented her from having the worst outcomes of this experience. The availability and her awareness of the counselling on offer was also key to her getting that support. Whilst the death of her son was an external factor that brought about grief, this alone did not make her seek counselling. Her innate desire to protect her children by doing everything to keep herself psychologically stable also drove this. This autonomous decision allowed her to be more receptive to the support on offer as well as the skill of the counsellor in providing a non-judgmental environment within which May could be open and honest (La Guardia, 2017). May's thoughts on her children not availing of the services at the time appeared to be accurate as she herself knew that you must be willing and ready yourself for sessions to be effective as it was for her other son, Ray.

Indeed, bereavement was what most participants thought would need formal support but only if individually or as a family you were not able to cope.

> *Nothing was offered, but I probably wouldn't have wanted any (psychological support). I think I felt that just doing bits and pieces myself was enough. I didn't feel like it was affecting me too bad. I think when you're in that bubble, yes, it's really hard. (Ben, ARVC, unclear screen)*

Many participants expressed that more support would be helpful, and this was suggested to come in many forms like having more CGNs, dedicated opportunities to talk through lifestyle recommendations and medical management with clinicians and

peer support which will be discussed in the next section. However, when asked directly about what their thoughts were about receiving psychological support from a clinical psychologist, there were mixed reactions.

Some differentiated themselves from those who needed formal psychological support by their ability to 'get on with their lives':

> *We just get on with our lives like normal. You get some people, like people who are unemployed, they're probably just sitting at home and they're thinking and dwelling on their condition because they haven't got anything to distract them. They probably need that sort of service more than us really. (Jo, F3, ARVC, pos screen & PGT)*

'Being stuck' and having a very serious form of the ICC were considered criteria for needing formal psychological support as well as being a type of person who would be receptive to this type of therapy. Linda and Jane offered contrasting views on the provision of psychological support. Linda preferred it to be a personal choice and the therapist should be competent and confident in providing this support whilst Jane felt that by packaging a psychological consult within the ICC pathway, this would facilitate a thorough psychological assessment and provision of services when required.

> *I think it would be a more personal thing if you needed that kind of help. You could approach somebody that is confident. (Linda, F4, DCM, pos screen)*

> *I think somebody that might not admit it, so would you run it as part of the package? Like, "Just go and see this person," then they're the specialists, aren't they? They can analyse the person and they know whether they need the help or not. I think you should make people do it. Maybe not give them an option, just say, "The next step is to see someone." (Jane, F4, DCM, pos screen)*

The preceding excerpts revealed that there was still stigma associated with verbalising the need and receiving formal psychological support (O'Donovan et al., 2020). Consultation with a clinical psychologist conferred a more serious state of physical and mental health issues which the participants did not identify with, even for most participants who had been bereaved or required more invasive management with an ICD.

May was able to recognise her profound grief and made the autonomous decision to seek help. It may therefore be the case that patients may have high levels of anxiety and distress but may not be aware of this and could result in delays in getting prompt psychological interventions. Jane's suggestion for psychological services to be incorporated into routine clinical care seeks to normalise the experience of seeing a professional. Whilst she used strong words such as 'not giving them an option', her emphasis on this consultation as an assessment helps introduce the role of the clinical psychologist in a non-threatening manner and a recognition that patients could benefit from having their psychological needs addressed but not all patients will need this more in depth support (Matthews et al., 2002)

Whilst most participants felt that they did not need any formal psychological support, most concurred that if they did need it, it would have been most helpful at the start of their screening/PGT journey or when they received their results which gives further credence to Jane's suggestion for an assessment presumably at an early stage to ensure prompt support when it is needed most. By framing it in this manner, a sense of choice is still facilitated despite the psychological assessment being part of routine care as patients should still have the opportunity to discuss how they would want to take matters further depending on their results.

6.3.4 Theme 4: Relatedness in the social context of Inherited cardiac conditions

The accounts of the participants demonstrated that events and actions related to being an at-risk family member for ICCs occur within a social context. By virtue of the nature of ICCs, this will always involve the family, this research has given more insight into the wider social milieu of ICCs. This theme deals with exploring the home and family, the ICC clinical service and other possible sources of support and how the participants' degree of relatedness with them can support or thwart perceived competence and autonomous decision-making.

6.3.4.1 Home is where the heart education is-coaching within families

By far, the most important focus for giving and receiving support for dealing with the risk for ICCs for many participants was their family. The importance of relatedness, particularly amongst the immediate family, was an important factor for participants in gaining skills, knowledge, and moral support to cope with their condition. For some participants who were parents, physically being with their children, even if they were already adults, during consultations helped ease them into the screening process (May, Mary, Linda, Val). The adult children welcomed this and did not feel that they were being undermined.

> *Although mum you've been up there a million times. And it's better with two of you there, because you take a bit more of it in. If it's just one person, then who do you talk to? (Bill, F4, DCM, pos screen)*

> *Yes, it's brought you closer together, I think. Well, we all compare our notes together, don't we? We all confide in each other. (Linda, F4, DCM, pos screen)*

There is an appreciation that clinical consultations may reveal bad news, convey complicated information so a second person, particularly one who is experienced with attending appointments, would be helpful in recalling and understanding the discussions. Bill also mentioned the long waiting times during these appointments and having someone to talk to perhaps ease anxiety and pass the time on the day was equally important.

There was a strong belief that partners, or a significant other should be able to attend the clinic so they know more about the condition and can respond accordingly in an emergency (Pam, Jo). As the family members went through screening, each had their own unique experience and reported back to the family to compare notes. Through this, they reassured, learned from, and gave each other advice; and both those diagnosed and those with normal screening results contributed. As well as comparing their symptoms, families also guided each other on how to navigate the processes in the clinic.

> *I used to get a lot. I used to feel like my heart was trying to jump out my chest. (Tina, F4, DCM, neg screen)*
>
> *I never get any of that. (Bill, F4, DCM, pos screen)*
>
> *I get that. It's always when I sit down. It was always when EastEnders is on. (Jane, F4, DCM, pos screen)*
>
> *What, when it goes "dum dum dum"? (laughter) (Tina, F4, DCM, neg screen)*
>
> *That's when I noticed it, because I think it might be a bit of, like that anxiety where you take a breath, and you feel like your heart's stopping for a bit? But I wouldn't know if it's just the way I'm sitting there relaxed and watching the telly. But I'd get it probably once a night. (Jane, F4, DCM, pos screen)*

What's the situation with my tablets? What do I have to do? Is it I've got to go to my doctor's? (Bill, F4, DCM, pos screen)

You've got to go to your doctor's (the GP), (the ICC consultant), he should have emailed your doctor with a letter. (Linda, F4, DCM, pos screen)

So I've got to make the appointment? They're not going to contact me? (Bill, F4, DCM, pos screen)

Yes. No, they won't contact you. (Linda, F4, DCM, pos screen)

If Linda had not known what the process was of getting a new prescription dispensed via the GP, Bill may have been waiting for someone to contact him and not be so proactive. This also highlighted some gaps in communicating the clinical pathway where some of the steps were not made clear by the clinical team and thus can cause delays in treatment.

This type of cohesion within families is favoured by several conditions: living and working close to each other (Linda runs the family business from home where all her children are involved), open and honest communication; and trust in each other. The shared experience of the sudden death in the proband has also strengthened these ties and common resolve to avoid the same outcome for another family member, and this trickled down to the younger members of the family.

Everyone was here the day that my sister (died), that it all happened. I think because we were all here, we've all seen it, we were all up the hospital. They don't need preparing, they're not sheltered children. They know what's going on. They know the

problems we've got, we're very open. They're tough, they just get on with it, like we do. (Jane, F4, DCM, pos screen)

A family theme seemed to have developed over this shared experience which influenced the way they dealt with change and stress (Galvin, 2019). In Jane's family, it is 'toughness' but with a scaffold of genuine care and concern. Participants, mainly the mothers, in the family group discussions have the tendency to speak on behalf of the other members like Jane has done, particularly along the lines of 'we are doing fine/well/getting on, aren't we?' (Linda, Val, May and Mary) which falls along the role of women tending to have the responsibility for health-related matters and more open to discussions around genetic conditions (Gaff et al., 2005). This statement was usually said with a visual scan around the room to check for agreement with the rest of the group members.

Group discussions were relaxed and there were no indications other members could not speak up if they did not agree with what was being said. Random questions and observations were posed without fear and the group members either tried to give answers or give their views on the situation freely. This respect for each other's perspective within these interactions supported one another's sense of autonomy as well as reinforce the sense of closeness and relatedness amongst the group (Ryan and Deci, 2017).

Families that are said to be functioning well are thought to be able to adapt to 1) the developmental stages over the family's life course and 2) the stresses and changing contexts they encounter (Gaff and Bylund, 2010). Over the years, some participants got married and had children and this excerpt from Julie showed how the additional role of affected mother to a carrier child was supported by the experiences of her sister, Hannah, who was the proband.

I think because I know how the girls (my nieces) have dealt with it, how it's been dealt with when they've been at school and they've got care plans in place and as Hannah said, "knowledge is power". We are aware of it, we know what to do if something happens and we know all about the drugs to avoid and kind of doing everything to make sure that we stay as safe as we can, you know, I think if I'd been in Hannah's situation and I had to learn that with my daughter growing up, then it would have been a lot different, yes. (Julie, F1, LQTS, pos screen & PGT)

Collectively, the family continued to increase their knowledge on how to deal with the condition and worked as a team to ensure a safe environment for themselves and the younger members of the family who were also genetic carriers.

And what we do as well, you know, don't hide in cupboards and jump out on the girls at Halloween because that could be catastrophic, but it is something we are aware of; it does help knowing that we've got the condition. (Fred, F1, LQTS, pos screen & PGT)

Solidarity within families at the time of the interviews was already strong, particularly for those who have experienced a sudden death, cardiac arrest, or extreme symptoms of ICCs but even more so in Julie's family where everyone who has been tested so far, apart from their mother, Val, has been found to be a carrier for LQTS albeit asymptomatic apart from the proband. As the family acquired new members, in particular, partners, they were open about the condition and shared information readily to ensure they were also aware of the precautions required.

Yes, I would say so, and all three of our children have got married since Hannah and since our family were diagnosed, and I think

initially we go in-depth to explain to you what Long QT is and how it's affected the family. (Val, F1, LQTS, neg screen & PGT)

Julie shared how her husband has prepared for caring for their own newborn, who is carrier for LQTS, by babysitting for her nieces is who were also LQTS carriers.

When Hannah's children stayed with us, or we've gone out for the day, he'll (my husband) always make sure that we've got May's defibrillator with us, and he knows all about that as well. And he's had CPR training and– because he's only ever had adult CPR training, and he's looking to get... (paediatric life support training). (Julie, F1, LQTS, pos screen & PGT)

Julie's brother had a new partner and shared how his partner has taken on board the seriousness of the condition and communicated with the family to support Julie's brother in terms of LQTS management.

I think she helped my brother take it more seriously because…I think my brother was a bit more blasé and like, "Oh I've got to take these tablets, and nothing is really wrong, and nothing is going to happen to me." And I think his partner has come along, she has done her own research and spoken to all of us and actually has made him realise that no, you do need to do it (take medications). (Julie, F1, LQTS, pos screen & PGT)

This responsiveness from new members of the family strengthened their relatedness as the care and attention they gave to preserve what the family values in terms of the ICC, in turn, made others feel cared for and related to and this was interpreted by the family as volitional giving rather than compliance to any pressure. The family's gratitude and relatedness would be undermined if they had sensed that actions from the partners were not autonomously motivated (Ryan and Deci, 2017).

There is a danger in families where family cohesion is too strong, and members lose their individuality. This is described as ‘enmeshed’ (Olson, 2000). This could result in isolation for any dissenters and a breakdown in family communication and relationships. To a degree, this has happened in Julie’s family with her brother’s ex-wife who ran counter to the family norm and decided to wait for her child to have PGT as discussed in section 6.2.2.3. Whilst they still have not come to terms with her decision and are very uncomfortable with the uncertainty, the family have continued to communicate as they value the relationship they have with their granddaughter and just take the necessary precautions.

> *I mean we see my son's daughter regularly, but the relationship with our ex-daughter in-law, has got better. But we still wouldn’t be able to convince her, even though we know when Hannah was diagnosed, we found out as much information as we could. And even to the extent that we just recently had CPR refresher courses, you know, we cover that side of it as much as we can. But yes, it is difficult. (Val, F1, LQTS, neg screen & PGT)*

Family members do not necessarily have to have the same diagnosis to be able to provide support and help with building knowledge and skills in those with an ICC in the family. As Paul proved, his experience with having an unrelated heart rhythm condition helped both his wife, Pat, who was eventually found to be a carrier for ARVC and his daughter, Rachel, who was the proband.

> *Like the first time I had a panic attack, which was brought on because I was getting…missed beats, and I could feel this thing and I didn’t understand why because I hadn’t had any... He was like, “Yes, you’re missing some beats,” and then I was like, “I can’t see, I can’t feel my arms,” and then I started screaming. And he was like, “You’re having a panic attack, calm down.” He took the day off work, which wasn’t much hard for him to do I have to say*

> *(laughter) to look after me and just to put my mind at ease. The great thing is, for me, that he's been through the worst. So when I say, "I'm scared, I'm really anxious," I know he knows what I'm talking about…We're quite lucky in that we can kind of understand how each other is feeling (Pat, F6, ARVC, pos screen & PGT)*

Paul can be empathetic with Pat and Rachel because he has experienced the same emotions and symptoms himself even if he did not have the same condition. He was aware of the degree of support and reassurance required to help others to come out of anxiety that is spiralling due to worry about palpitations as this can get worse if the individual is not able to relax and calm down. Both Pat and Rachel have seen him successfully cope with this and therefore have the confidence to approach and trust him to gain reassurance and he willingly gave his time for this.

Paul and Pat laugh about his 'taking a day off' as it has been mentioned before how Paul was very much occupied with his sport but on matters such as this for his family, he takes the time to be there for them. Central to autonomy support in parenting is empathy to enable appreciation of the child's point of view. In helping to manage symptoms like palpitations in ICCs, Paul can empathise with his child both on a physical and emotional level and this also applied to his wife, thus, strengthening the degree of relatedness amongst them within this shared experience. It could be argued that their kinship ties made it more natural and easier for Paul as a parent and husband to give this support, but we have observed in this research how issues around ICCs can also adversely affect relatedness, particularly amongst couples.

As parents get older, caring responsibilities start to shift to the younger generation, highlighting their increased awareness that elderly parents are becoming frailer and need support. Sheila was in her 80s and her son, Peter, who has HCM, and special needs was in his 50s. They moved to the countryside to live together in a compound

with her other son, Bob, so that they can have additional support. The family have lived with HCM for several years and have learned to deal with the practical aspects of looking after Peter. Now that Sheila is getting older, Bob appeared to be trying to connect with Peter more on an emotional level with the HCM. In part, to prepare himself to eventually take on more responsibility for caring for him.

> *When he went in and had his defibrillator fitted, Peter is Peter. You look at him and you don't really see a massive change in his lifestyle. I don't know what goes through his head…He obviously does think certain things and he doesn't really know how to complain about something. I know it's a different aspect, being that I've had a heart attack; I know you don't carry on the same way as you did before. That makes me think a little bit about him. Does it affect him? Has it affected him? Very difficult to say.*
>
> *What she (Sheila) has got there with Peter with his ailments, his defibrillator, it's her problem, her issues, her child. She will deal with that until she dies. But she can't cope with it anymore. I look at her and think, "What happened to my mum? She is getting old." No kid wants to admit that.*
>
> *If something happens to me, what happens to them? …Because I need to be here to look after them and make sure they're all right, make sure no one takes the mickey out of them, make sure that they can cope. (Bob, F2, HCM, unclear screen)*

Bob is getting ready to take on a bigger role in Peter's care as Sheila gets frailer and although he has learned how to care for Peter's physical needs through the years, he realised that he must deepen his relatedness with Peter on an emotional level to enable him to understand and care for him better. He has used his own experience with his prior possible diagnosis of HCM and the subsequent heart attack as touchpoints for what he would like to know more about Peter. Bob is taking the cue

from Sheila as she is the authority on Peter's care and during the interview, she was starting to open up to accepting this support.

Learning about how to live with HCM has two aspects in this family. The first is working together to look after a vulnerable family member with HCM, educating each other on the practical aspects of the disease; and the second is having a deeper understanding of the emotional aspects of the condition for the affected relative. In some families, the two aspects occur contemporaneously and whilst to some degree, it is probably the same for Bob, the second aspect becomes more of an acute learning need for him after his own health issues and recognising the increased frailty of his mother. Therefore, to enable families to function optimally when caring responsibilities are shifting, they acquire flexibility in decision-making roles and allow negotiation (Olson, 2000) as this opens up the opportunities for the younger generation to learn more from their elders.

Lisa also had many relatives who were diagnosed with Brugada Syndrome, and her own father died suddenly of the condition at a young age. She was the only child from her father's partnership with her mother and relationships with her paternal side of the family have not always been smooth. Therefore, communication amongst the family was not as open as she would have liked it to be, and this has proven detrimental to her psychological need for relatedness and belonging.

> *Two of my sisters have got it (Brugada Syndrome). One of my brothers has got it, my other brother won't have screening, and my other brother died. And a lot of their children have got it. I think I am the only one at the moment that's got the baby with it. So I don't hear a lot from them and that's what makes me angry, because it's just me. It's just me and (the CGN) and (the cardiologist) It's no one, is it?...And that's what frustrates me, to think that I did it all on my own, when I could have had them*

supporting me a bit. But here we are today seven years down the line, and we're still going strong. I can't do more than be a good mum and be there for Matthew. (Lisa, BrS, pos screen & PGT)

Lisa experienced the opposite of the other participants and instead of feeling a greater sense of relatedness, she felt more alone in terms of her ICC experience as there was no one in the immediate household she could relate to as none of them were at-risk relatives. However, she found support in her partner who was with her from the very start of her patient journey.

That was really hard because I thought, "My dad isn't here anymore. The only person is me, because my mum hasn't got it…And the only person I have is my partner…He's been really good. (Lisa, BrS, pos screen & PGT)

As the interview continued, Lisa started to recall some of her relatives who she could relate to in terms of the ICC and although she was glad to have someone with her when she got the results of the Ajmaline test, what was missing was the solidarity she was hoping for in someone who also had the same results.

When I found out about this I was sitting round my nephew's house, and my nephew, that's my brother that died, it's his son. And we both had the ajmalines at the same time and he didn't have it. And that was a real shitter on me. I know it's horrible to say that but at least I had someone to be with…So I've got this group from the boys, which is really good because a couple of them are younger than me. That's the only, really – and my uncle in Australia, and my Auntie Anne…then her daughter's got it. There's a lot of us, yes. (Lisa, BrS, pos screen & PGT)

Although Lisa's family by far has the largest number of people known to be affected with an ICC, it was very stark that this has not become a network for her to gain a

greater sense of relatedness through the condition. This supported perceived competence for living with the condition by the other family group participants. This demonstrated the extreme end of family cohesion in terms of her relationship with her extended family which could be described as ‘disengaged’ where there is emotional separateness (Olson, 2000).

Lisa’s basic psychological need for relatedness has been thwarted by this situation and has resulted in feelings of frustration and resentment. This could have resulted in ill-being (Ryan and Deci, 2017), however, this lack of belonging in one group made her appreciate the support of her partner more and strengthened her resolve towards personal growth, health and a focus on raising her son. In the future, this need for relatedness in the context of the ICC may be fulfilled as her son matures as this is something they share.

Relatedness in the social context of ICCs can be deepened and strengthened in the family unit. This is achieved as family members coach and support each other and have shared experiences as they go through the screening or PGT process and long-term as they live with the condition.

6.3.4.2 The clinical service and health professionals in inherited cardiac conditions-scoops you up

The clinic set-up and the team of healthcare providers that support at-risk family members as they go through cardiac screening/PGT was another context for participants that impacted on how they were able to cope with the process and their results. The perceived quality of relationships and interactions participants felt with their clinical team either satisfied or thwarted their basic psychological need for relatedness.

The support that the NHS provides was fundamental to participants being able to cope with how the ICC affects them and their families. The confidence in having services available without having to worry about financial costs removed what could be a very significant issue for those who do not have the benefit of a state-funded health system.

> *The care that I've received? Ten out of ten, yes. No issues with that whatsoever, I'm so grateful to be in the National Health system with this, I can't tell you. My sister in (the Republic of Ireland) with twins, as a single mum, no support there at all. That's expensive and frightening. So no, I'm extremely grateful for that. (Karen, LQTS, pos PGT)*

Within NHS, the other care system that provided a network of support and relatedness in the social context of ICCs is the specialist clinical service. In its totality, it is described as system which provided the process of screening and/or PGT and the aftercare required. In addition, it was perceived as a comprehensive resource where participants and their families can turn to for all their needs associated with coping with the ICC:

> *So it was horrific but then we felt so lucky at the same time. Then you get on this journey, don't you? With the (ICC service in the hospital) and everybody, and we just got scooped up. So it was definitely a mixture of being really frightened actually, but also feeling lucky that you're in amazing hands. (Pat, F6, ARVC, pos screen & PGT)*

Despite a life-changing diagnosis, participants still felt hope and optimism with the knowledge that there was a corresponding specialist service to support them in all aspects of the condition. There were many features of the service that contributed to this perception. The set up and flow of services as described in by the participants

fulfilled the basic criteria for accessibility and general efficiency. The characteristics of the clinical team were also important factors, this included expertise, the willingness to listen and answer questions as well as participants felt they were given adequate time for their consultation; not rushed or pressured into a decision.

> *And I felt that you all had as much time for me as I needed. I'm sure I asked loads of very dumb, simple questions and I'm pretty sure I asked some of them more than once. And (they) were so accommodating of that. For me it really was just a fact-finding mission, and you guys delivered on that perfectly. There was nothing more I expected or wanted from that. So I was very happy…I guess then what you would say is that what (the ICC team) have offered to me has been the most important, for me to be able to make that informed decision myself. So who's been the most supportive? It's actually been people (like the ICC team), helping me steer my way through all these decisions that I make myself…I really mean that. (James, BrS, dec screening)*

James, as previously mentioned, did not go ahead with screening at the time of his visit. His excerpt confirmed the autonomy supportive environment that the ICC provided and that he felt respected rather than judged when he made the decision not to go ahead with screening. This was not without the reassurance that he can access the service should he change his mind and indeed, he eventually approached a local ICC service where he moved when he needed them. A more paternalistic attitude to his care and/or lack of understanding behind his decision-making on the part of the ICC team could have resulted in James disengaging from ICC services which would have been more of a risk to his health as he was eventually diagnosed with BrS.

Whilst participants spoke of their relationship with the ICC team, some also singled out the excellent care and regard they had from specific members of the team. The

CGN role was frequently mentioned as someone who was supportive, easily accessible, and responsive (Lisa).

> *The only thing I'd like to say is that I'm just really grateful for the support we get from (the CGN) mostly, (she's) always at the end of the phone. I know when I spoke to (her), just before I had Rose and then just after, (she) put my mind at ease straight away, and I think that's why we're able to take everything in our stride, because of the support we get from the (the ICC service) and specifically the CGN. (She has) been incredible for our family. Always there and response times are phenomenal. (Julie, F1, LQTS, pos screen & PGT)*

The CGN going above and beyond her working hours to help this family also demonstrated that relatedness worked both ways in that there was a deepening of empathy for the concerns of the family and whilst the CGN's actions stayed within professional boundaries, there was a recognition that some concerns needed to be addressed more urgently. This personalised approach and the ability to offer practical help and advice made it easier for participants and families to gradually gain perceived competence in coping with their condition followed by developing confidence to access the wider ICC team for specific issues.

Part of the wider ICC team is the high-risk pregnancy team. As patients affected by ICCs can be of child-bearing age, having these allied teams easily accessible and coordinated within the ICC clinic provided seamless care for participants who were worried about their own health as they carried a pregnancy and that of the foetus. Julie shared her reflections on her experience of perinatal care:

> *I don't think I've ever seen as many doctors in one room, sitting down. But very, very knowledgeable, put my mind at rest straight*

away, because it was a concern with pregnancy. They were incredible, and they said, "Look, if you need any assistance with your local hospital where you have the baby, just come back to us. Or if you think of anything." So it was very much, the door was open to go and see them again if we needed to. And then when I told (the ICC consultant) I was pregnant, he said, "Right, we need to see you in clinic a bit more just to keep an eye on you and specifically afterwards as well." So that's all been done, and I think when I saw the anaesthetist at my local hospital, I think they even got in touch with (the ICC team) – because they'd never really come across Long QT in pregnancy. (Julie, F1, LQTS, pos screen & PGT)

This network of care to support a pregnancy that spanned geographical regions and involved a multi-disciplinary team, further strengthened the relatedness of the family with the ICC service. However, at times, even simple clinic consultations can go wrong, and this can be a threat to strong relationships built over the years.

I think I've seen (the ICC consultant) never more than two or three times. But I know that he doesn't need to see me, he's a very busy man. I don't have an issue with that. It's just an issue with my information. Sometimes you go in and you wait a long time for the appointment and then they don't have the results. You go, 'But I had that monitor on six months ago." It is a bit unsatisfactory sometimes in that, because you only have one slot to see a doctor once a year, and you have to make a big decision: "Do I opt for Beta Blockers or do I need to be doing something else?" But the information you need to make the decision is not there for the doctor to help you.

I said, "How do I get my records from here to there?". (The doctor) left me to do that and I hadn't a clue how to do it. Just really the thing about joining up the dots about how to work inside the system, how to – you know, if the doctor says one thing inside the

> *surgery but then it never gets into the computer. You're sitting there going, "How do I make that happen?". (Karen, LQTS, pos PGT)*

The exasperation in Karen's excerpt was quite clear as she experienced the breakdown in communication amongst the ICC team. This resulted in her not being able to make the most of her time with the ICC clinician in making an informed decision as neither of them had the necessary information to hand. Therefore, inasmuch as trust and relatedness can be strengthened by executing complex care plans as in Julie's case, this can also easily break down by a simple lack of communication in a single clinic visit.

Yet, Karen, like many participants, trusted the expertise of the ICC consultants and this was instrumental in participants having confidence in their medical care as they felt they were in 'good hands', especially if the consultant had diagnosed and seen the proband through an acute episode (Mary). This sense of relatedness became stronger when, over the years, a more personalised approach was taken by the ICC consultant which has led to better relations particularly amongst those who had affected/carrier children and their paediatric ICC consultants.

> *The man's (paediatric ICC consultant) incredible, he's been great and the girls love him, and I feel very reassured by him. And he interestingly has changed his approach to us as well; I don't know what that's about, but he's kind of taken more our approach to their care. (Karen, LQTS, pos PGT)*

However, as families build their relationships with their clinicians, there was a growing fear that if the doctors retired or left, they would lose the trusted relationship and expertise that they received and the reassurance they have is only temporary.

> *And I think because he's so knowledgeable about Long QT, like he is one of the best in the country, and I think that's another worry isn't it? Is whoever takes over from him going to be as good and as thorough? (Val, F1, LQTS, neg screen & PGT)*

Only Lisa credited the role of the local cardiologist who implanted her ICD as 'supporting her all the way'. Indeed, some participants intertwined the quality of their care with the ICC consultants and the ICC service so much that they found it hard to trust other clinicians and local services.

> *Yes, that's the other thing. I wouldn't want to go anywhere but (this ICC centre) and under (my ICC consultant) and (the team) because they know of us for ten years now. Yes, you feel comfortable going there. You know that they know you, they know your condition, they know your symptoms and they know your history. (Pam, F3, ARVC, pos screen & PGT)*

The attitude of the participants towards the specialist clinics are reinforced by published guidelines in ICC care which state that patients and families should be seen in such clinics as the gold standard (Ackerman et al., 2011, Elliott et al., 2014). Furthermore, in the systematic review in Chapter 3, we saw how patients and their families were frustrated and may have come to harm because of a lack of expertise in the early days of ICC management. As trust and good working relationships builds over the years amongst patients, families and clinicians, a sense of relatedness brings confidence and the ability to cope with the impact of the ICCs as patients know that the specialist clinic continues to monitor them and there is easy access.

However, as health services on a whole evolve, relatedness in the specialist services must not come at the cost of building trust and relatedness with other services,

particularly primary care services and local hospitals (most ICC services are based in tertiary centres) as ICC care sometimes depend on local services, especially if patients have concerns about traveling and time off school and work (Tom, Ben, Steven, Tim). GPs are also essential to providing patients with comprehensive care, not just for ICCs. This is difficult when patients felt let down or have no faith in their local systems.

> *That is a problem, that I really don't have much faith in our GP. Never have done. That was our first port of call and they got something wrong. For example Rachel, every now and then she gets ulcers in her throat. Which we're thinking is probably a side effect from the nadolol. But it's doing such a great job on her rhythm, we think, "Well she's probably going to have to put up with it." I would like for her to go to the GP and say, "Is there anything you can do about this?" But I don't feel like I'd be confident. So we'll probably have to wait and see (the paediatric ICC consultant). (Pat, F6, ARVC, pos screen & PGT)*

> *So is it somewhere you can speak to someone and say, "Right, my child has now hit 30, what should I be looking for, because in the family we have this?"…Or the other thing is, my child has hit 13, can I come in to have them screened, just to check, rather than going through your GP who goes, "Oh no, go away, come back in a couple of weeks if you're still worried." (May, F5, HCM, neg screen)*

May was keen for a service where family members are able to get screening without having to go through the GP. Indeed, many ICC services are allowing self-referrals for family members at risk for ICCs to decrease the delay in referrals and burden to the GP. However, in areas where this system is not in place, individuals like May would be less averse if they felt that the GP would be receptive and act on their concerns promptly. This hesitation from May and Pat to approach the GP stemmed

from a lack of relatedness and may cause a delay in care especially if they have not established strong links with the ICC service.

6.3.4.3 The listening ear of friends, employers, and patient support groups

The social context for families affected by ICCs also extend to friends, employers, and patient support groups. Examples of how these three social groups have been helpful in supporting participants through their patient journey were given and highlighted the potential for enhancing these relationships to increase the satisfaction of the need for relatedness.

For Linda, her diagnostic journey was heralded by the observation of worrying patterns of sudden death and prompting by a friend.

> *Then everything started to fall into place after that. Because, at the time of my brother's death in 1991, one of my close friends said to me, "I'm really worried about you, I want you to get screened," because I was living on my own with just the two girls. She said, "If anything happens to you, I want to make sure you're okay." (Linda, F4, DCM, pos screen)*

In times where there is no other adult family member as a source of support as in Linda's case many years ago, a close friend may be the only person who can help gain perspective for a risk for an ICC. This clearly had a huge impact on Linda as she recalled the key role her friend played in supporting her health even after many years have passed.

Friends and peers also play a huge role in supporting young people, whether it is as a group to keep an eye out for one of them who has an ICC or providing emotional

support. Two siblings described how their peer group helped them in both these situations:

> *I think I told my friends because I think at the time it was quite upsetting. I also didn't want to show that around Henry (brother who was the proband) either, because I think we were all quite uncertain about what was going to happen. (Sharon, F7, neg screen & pos PGT)*

> *No one really talks about it, I suppose. I think everyone is a little bit aware of it, I'd say more the boys probably than the girls. More Henry's friends, of if I'd say something they would possibly be thinking the same thing as me. (Kate, F7, neg screen & PGT)*

Much emphasis was given by the siblings of not discussing their stories in detail with their friends and yet, the peer group was described as 'having an understanding' of the situation, supportive and can be called upon to help in case the proband fell ill during their outings.

People are generally selective regarding the person they will turn to for emotional support and this is usually predicted by their perception of this person's autonomy supportiveness (Lynch et al., 2009) and ability to take on their internal frame of reference (Deci and Ryan, 2014). The relationship of the siblings with their peers attested to this as they did not feel pressured to reveal more than they have already discussed about their situation regarding the ICC yet felt secure in the group's support. This volitional emotional reliance towards their peers displayed by this group of participants is thought to bring about greater basic needs satisfaction, particularly for both autonomy and relatedness, resulting in greater psychological wellness (Ryan and Deci, 2017).

Participants who were diagnosed or found to be genetic carriers for an ICC all remained in employment at the time of the interviews. For those employed, the workplace was an important social context as even the cardiac screening and monitoring pathway requires several visits to the hospital.

> *My boss at work is, I won't say fully aware, she knows that I have a heart condition and that it has these risks associated with it and I need to visit hospital this frequently. She is incredibly supportive and she would never hold me back from taking time off work to go and visit my cardiologist if I needed it every six months or every two months or every two weeks…so it's not affecting my work in that way. (James, BrS, dec screening)*

Becoming more symptomatic with an ICC necessitated more modifications in the workplace for some participants.

> *If I'm at work I'll tell my manager if I'm not feeling well and at least he knows to come up and keep an eye on me every now and then, instead of letting me carry on doing whatever. Before I was lifting heavy barrels and stuff here and there and my manager doesn't let me do it now. He gets somebody else to lift all the heavy stuff. (Jo, F3, ARVC, pos screen & PGT)*

These participants felt secure in the awareness of their managers of their ICC and the support the mangers gave for periodic check-ups and adjustments to work duties. In James' case, he was able to communicate his situation in a way that did not breach his own threshold for confidentiality whilst for Jo, he more likely had to reveal more information to enable him to gain the necessary adjustments at work. This is not always easy for ICC patients due to the perceived risk of losing their jobs and the associated emotional and financial burden that ensues, particularly for those who

have manual jobs or operate heavy machinery/vehicles (Etchegary et al., 2016). Jo had a manual occupation, but his employer seemed to be accommodating of his needs.

Whilst joy and satisfaction in their work despite an ICC diagnosis was not the main point of discussion in these excerpts, there is an indication that this ability to have these conversations and a receptive and understanding manager, provided needs satisfaction for relatedness and retained their motivation in their job roles. Gainful employment is an important factor in maintaining a sense of being valued and a positive outlook for the future life prospects in long-term conditions (Vassilev et al., 2014).

Whilst the workplace could provide a supportive environment by responding to the needs of an employee with an ICC, it may also facilitate emotional and psychological support depending on the nature of the business the company is involved in. Ben worked with a heart charity and by virtue of the specialist industry they are involved in, this has given him an unexpected social context from which relatedness needs could be satisfied.

> *Well, working for the (heart charity), it's hard to get away from it. And it's nice when I'm at the shop and I'm on the till and I hear people's stories and what they've been through, and again, it puts things into perspective. They've had open heart surgery and stents put in and all these different ways they've survived heart disease. I think that's helped as well…it's like little mini support groups at the shop, yes…I get people saying, "I'm here today bringing donations because I lost someone," or "Because my dad's got heart disease." …And then I'd mention my sister sometimes, if I felt it was appropriate. Yes, it's just nice to open up. (Ben, ARVC, unclear screen)*

Through Ben's conversations and sharing stories with those affected by heart conditions, he has formed connections and experienced relatedness that reduced his sense of isolation as we previously learned how hard it was for him to share is possible diagnosis of ARVC with his parents. Another important aspect of this social context was that whilst he has nothing in common in terms of the ICC with these heart patients, their range of experiences with clinical services gave him a perspective on his own relatively non-invasive cardiac interventions resulting in a more positive outlook. These effects are not dissimilar to those observed in structured peer support programmes in chronic diseases (Embuldeniya et al., 2013).

Ben's customers at the heart charity shop created an informal support network for him but there were also formally organised patient support groups that some participants sought. Whilst the main aim in signposting families to support groups is for them to be supported, participants describe how they were also able to give back via the support group.

> *We actually have helped SADS (Sudden Adult Death Syndrome UK) because they helped us initially, very much so, you know, put us in touch with other families who had gone through the process and come out the other side. And we've since done the same thing… (Julie, F1, LQTS, pos screen & PGT)*

> *SADS helped us out when Hannah was diagnosed, and they helped fundraise to get a defibrillator for May's nursery…Dan did the London Marathon this year, another friend done – even though they weren't part of our family when Hannah had her cardiac arrest, they've seen how much SADS have helped us, and as a family we like to give back what we got out. Because it helped us immensely…even down to the leaflets. (Val, F1, LQTS, neg screen & PGT)*

Peer support provided by a patient group like SADS UK usually involves emotional informational and appraisal support (Dennis, 2003) and from Julie and Val's excerpt, this was what they gained alongside practical support to fund an external defibrillator for the school of a young affected family member. This network has extended Julie & Val's family's source of relatedness satisfaction beyond their immediate family and friends. 'Having come through the other side' , Val and Julie's family have started to give back to this social network they perceived to have helped them adjust to and cope with their ICC diagnosis.

Indeed, in SDT, it is postulated that people find inherent satisfaction in helping non-kin others (prosociality) and in doing so, facilitate the satisfaction all three basic psychological needs and benefit from a sense of vitality and well-being (Weinstein and Ryan, 2010). In this particular family, competence was experienced as they effectively helped others through sharing their own experiences and participating in fundraising; relatedness was further enhanced by developing a sense of empathy and interest in others; and as the family's actions were unforced, autonomy was practiced.

The profound sense of relatedness that Julie and Val felt towards the SADS patient group may have developed over time, but this can also come about within a short period. Lisa was in a group discussion with a parent-couple, Pat and Paul, and within the interview, had offered her support for them and their daughter, Rachel.

> *But like I say, when Rachel does have to have this thing done, obviously she will be in the best place, I'm more than happy to talk about it and tell you everything...If I can help someone, if I can put someone's mind at ease, I'd be happy. (Lisa, BrS, pos screen & PGT)*

Lisa was acting with autonomy in extending her offer of support of which she felt competent to provide to a couple where she established a sense of relatedness. Again, the human tendency for prosociality and helping others described by Weinstein and Ryan (2010) is evident in this excerpt. it was likely facilitated by Lisa identifying with the same attitudes for coping with the ICC situation and empathising with the couple and their daughter, who will soon undergo an ICD implantation which Lisa had previously mentioned as a frightening experience for her and where she did not have a lot of family support.

The social contexts in ICCs are broad and go beyond the immediate family. Fostering and satisfying the need for relatedness in participants through these networks was essential for ICC families to learn from each other in terms of coping and adjusting to the ICC; and feel secure in their medical care and employment. Furthermore, these networks also gave the participants an opportunity within which they can also give back and help others, which provides a healthy medium by which to satisfy all three basic psychological needs.

6.4 Findings directly related to the psychoeducational intervention design and recommendations from the participants

This study has also provided more insight into the possible modes of delivery of the psychoeducational intervention, including online, individual and group sessions. Group sessions appeared acceptable to most participants and gave the added benefit of learning from their peer group. However, it was also noted that sharing stories may also trigger anxiety, thus, careful facilitation of sessions is required.

The significant time periods described in this study: initial awareness of risk, attendance at an ICC clinic, receipt of results and the immediate period thereafter; and longer-term adjustment to the results; can all be considered for timing of the

intervention. However, the overwhelming need for support appears to be at diagnosis or receipt of results in the form of a formal programme akin to cardiac rehabilitation; with some ad hoc support during the follow up period. The advantage of the intervention being offered as part of routine care would then remove the stigma associated with the terms ‘psychological support’ which many participants felt was ‘not for them’.

Other considerations for the intervention included provisions for specific groups such as bereaved families who may need more time to talk about their grief and access to formal bereavement support; and parents of probands who may need to work through issues around their sick child before they are able to focus on their own health.

Apart from the components and features of the psychoeducational intervention derived from the analysis of the evidence generated by this study, participants also offered suggestions directly to inform any additional support to be offered to them and any improvements to the existing care they received. These are summarised in Table 10.

Table 10 Considerations for the psychoeducational intervention

Key content and implementation strategies	Analysis of qualitative data	Participants' direct suggestions
Content	**Supporting information needs:** • Cardinal content on inherited cardiac conditions (ICCs), including genetics • Management strategies (risk stratification, medications, symptom recognition) • Tailored lifestyle advice • ICC care pathway and access **Supporting psychological well-being:** • Communication skills • Building confidence • Problem solving strategies • Coping skills • Peer support	**Kate & Fred:** tailored to age groups

Key content and implementation strategies	Analysis of qualitative data	Participants' direct suggestions
Format and delivery mode	• One-to-one support from the healthcare team, likely to be the CGN • Facilitated group format mixed with peer support • Delivered within the family as a group or groups of families	**Mary, Bob, Fred, Linda, May & Jane:** Group meeting **Fred:** Facebook group **May, Pat & Jo:** Phone or email support **Louisa, Linda:** Written or online information **Kate:** YouTube videos **Pam & Linda:** Involve family
Access and duration	• Offered as part of the care pathway and available throughout long-term follow up	**Maria & Pat:** Something you can dip into **May:** Easy access phone line **Bob & Jane:** Part of prescribed programme

Key content and implementation strategies	Analysis of qualitative data	Participants' direct suggestions
Timing	• Some aspects more pertinent at certain time periods but emphasis on the immediate period after a diagnosis and/or predictive genetic test results	**Ben, Val, Bob, Pam & Jo:** At diagnosis or receipt of results
Implementation considerations	• Reliable, accurate and with flexibility for individual approach • Not labelled as 'psychological support' • Consideration for certain groups: Bereaved families may need specific input or time to discuss their grief • Parents may also require support to focus on their personal health	**Pam, Lisa & Linda:** Discussing their stories may increase anxiety

Chapter 7: Phase 2 Developing the intervention model- Qualitative study discussion

7.1 Introduction

The aim of this qualitative study was to gain the perspective of the intended end-users to inform the development of a psychoeducational intervention being developed for this research. This study has brought forth the experiences, views, and preferences of family members at risk for an ICC as they undergo cardiac screening and/or PGT. Four major themes have been generated from this study: Impact of the proband's story, Leveraged autonomy, Harnessing competence and Relatedness in the social context of ICCs. The themes come about against the backdrop of important timepoints and activities within the cardiac screening/PGT journey which commences at the initial awareness of risk, followed by attendance at an ICC clinic where screening and/or PGT was likely to occur in most cases; receipt of results and the immediate period thereafter; and finally, the longer-term adjustment to the results. Opportunities and targets for the psychoeducational intervention were also identified which will be described in Section 7.4)

7.2 Application of Self-determination theory as the theoretical framework

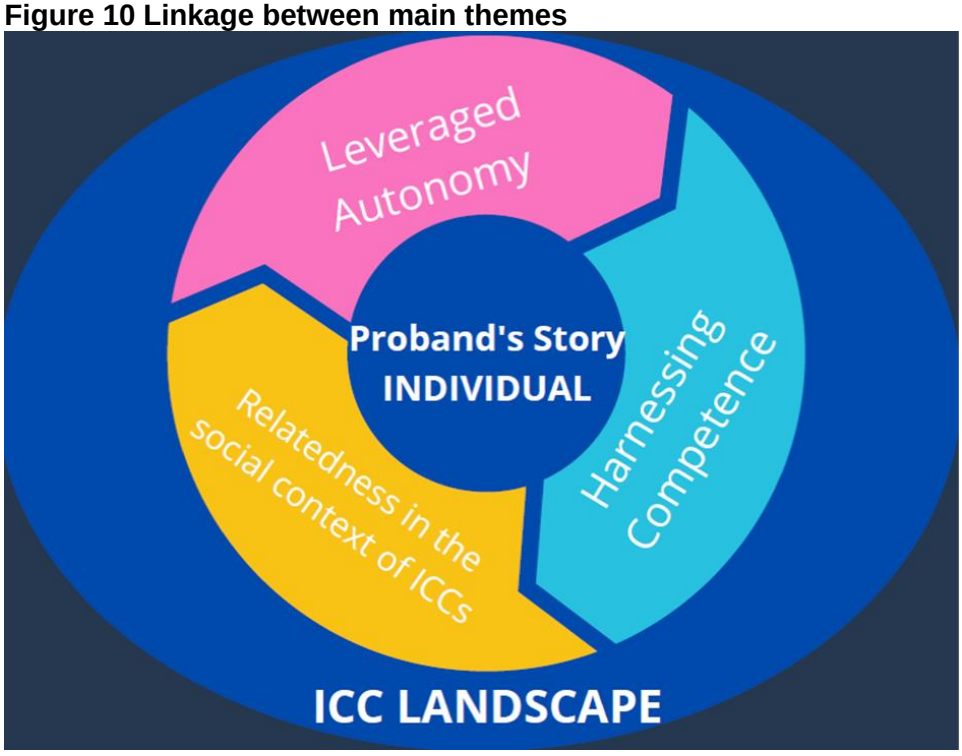
Self-determination theory was chosen as the theoretical framework to underpin the development of the psychoeducational intervention for this research as it is consistent with the family context of genetic conditions. The application of SDT in the analysis of the qualitative data generated by this study was implemented in the later, Explanatory phase of the analysis, and 'matched' to the data. Through this process, it was revealed that the basic psychological needs of Autonomy, Competence and Relatedness dominated individually within subthemes to enable a higher order main theme, as well

as all three needs present across themes. This has resulted in SDT providing a coherent thread in terms of analysing the views, experiences, and preferences of the participants; as well as providing explanations for participant motivations, actions, and outlooks.

7.3 Linkage between main themes

Figure 10 illustrates that the starting point for most of the participants in their involvement with ICCs is within the theme Impact of the proband's story, but all four main themes are interrelated. Whilst the other themes: Leveraged autonomy, Harnessing competence, Relatedness in the social context of ICCs herald the three basic psychological needs within SDT, these needs interact to help fulfil one another to support the individual to grow, adjust and thrive within the ICC landscape. The linkage between themes is explained in greater detail as each theme is discussed.

Figure 10 Linkage between main themes



7.3.1 Impact of the proband's story

The theme, Impact of the proband's story formed the basis of participants' narrative as they become an at-risk relative for an ICC. Sudden death or severe symptoms in a proband impacted the individual and whole family profoundly and whilst these events were external to the participants, thereby a source of extrinsic motivation for action, this study showed that the process of internalisation quickly ensued. Internalisation is described within SDT as a process whereby values, beliefs or behavioural regulations are taken in from external sources and are transformed into one's own (Ryan and Deci, 2017). Whilst intrinsic motivation is the highest form of self-determined behaviour, internalisation supports the most autonomous form of extrinsic motivation which is described as behaviours that have integrated regulation or self-regulated as opposed to the other end of the spectrum wherein behaviours are externally regulated (Ryan and Deci, 2000).

It is evident that over time, these internalisations became more integrated in participants through a continuous process of self-reflection and identification, not just with the proband but also their family and their values. An advantage of a high level of internalisation includes individuals experiencing their behaviour as more volitional, and therefore more autonomous. Furthermore, competence is satisfied as individuals feel more efficacious in their actions and as they were brought closer together by the proband's story, the need for relatedness with their family is fulfilled. Rejection may be a potential by-product of internalisation within groups because of differing beliefs (Ryan and Deci, 2000). The participants described family members who became estranged because of this but did not actively seek to sever ties with them.

Initially, when there was a death in the family due to an ICC or a diagnosis made, relatedness came into play for the participants as they rallied to support each other or the sick relative. This relatedness very quickly came into sharp relief with autonomy

as the next actions for them as an at-risk relative needed to be taken. The automaticity of actions for screening/PGT and apparent lack of opportunity to consider options amongst the participants were a threat to autonomy and could have led to post-decision regret (Payne et al., 2000).

In SDT's view, very few intentional actions are truly autonomous (Ryan and Deci, 2017), and this was also evidenced in this study. Even participants who were parents, who tend to be intrinsically motivated when taking action to protect their children, were constantly debating within themselves whether they were being coercive versus autonomy-supportive in their parenting as regard to ICCs. This discomfort with some of the decisions they have made is an indication of incongruence with the self and a threat to autonomy (Friedman, 2003).

7.3.2 Leveraged autonomy

In general, health behaviours are not intrinsically motivated as they are not done solely for inherent interest and enjoyment but are performed in the service of a goal (La Guardia, 2017), for example, prevention of sudden death in ICCs. Therefore, the significance of internalisation to achieve self-regulation of health behaviours is linked with the next main theme as this study demonstrated that most actions and decisions relating to cardiac screening and/or PGT were performed with Leveraged autonomy. These 'levers' included concerns for personal health and family relatedness which appeared widely internalised within the participants as generally, autonomy did not seem to be impinged.

Where autonomy seemed to be most threatened was in the screening and/or PGT of the younger participants and in a few older participants. Although none of these participants expressed regret, it was evident that the sense of choice and/or the completeness of information before deciding was not implicit.

This threat to autonomy comes hand in hand when infants or very young children have PGT as parents will ultimately make this choice. The general guideline for PGT is that young people should have the opportunity to have comprehensive genetic counselling to make an informed choice about whether to proceed with PGT or not (Clarke 1994). In those who have no capacity to consent, where results will inform their immediate diagnosis, treatment or surveillance, it is usually in the best interest of the child to offer PGT (RCP et al., 2019). In LQTS, such as in Family 1, PGT can be offered to young children as beta-blockers and the avoidance of prolonged QT inducing drugs are protective against dangerous arrhythmias so there was no conflict in terms of medical recommendations in this situation. When parents make this choice for their child to have PGT, it is therefore recommended that they should have genetic counselling support as age appropriate when they are older.

Satisfying the psychological need for competence in the aspects of informational needs as well as social competence therefore comes hand in hand with promoting autonomy as participants navigated the cardiac screening and/or PGT pathway, to enable them to challenge and ask questions when they were unsure.

7.3.3 Harnessing competence

The theme Harnessing competence highlighted further where more input is required to support the satisfaction of competence needs and what fostered or hindered participants in attaining this. In terms of informational needs, it was apparent that each significant time period along the cardiac screening and/or PGT pathway required its own specific input. When participants did not find this information easily, they would seek other sources that may not be reliable, wait until their next appointment or in the case of some of their relatives, become lost to follow-up. The latter outcome is a sign of amotivation, a state wherein one is not motivated to behave and lacks intentionality

and can easily occur due to a shortfall of perceived competence (Legault et al., 2006). This is where competence in terms of information is intertwined with social competence as both are required, particularly for those who are diagnosed or are genetic carriers, to ensure engagement in care pathways for long term conditions such as ICCs which require multiple clinical tests over periods of time; and the need to communicate risk to other members of the family.

7.3.4 Relatedness in the social context of Inherited cardiac conditions

It was evident within the Harnessing competence theme that participants behaved in the direction of coping, growth, and adjustment to their situation. The degree of relatedness they had with their family, and the relationship they were developing with the clinical team were emerging as key factors to help attain this. This is where SDT has shown its high applicability to genetic healthcare as the social environment and the need for relatedness within this, are given emphasis as a basic need for psychological well-being.

The last theme, Relatedness in the social context of ICCs, demonstrated how the participants' social environment can hinder or progress the ability to navigate the cardiac screening and/or PGT pathway. As families became more experienced with ICCs, more resources for information and support became available to the individual member. With each satisfactory clinical encounter, even if they received bad news, trust and confidence was being built between participants and clinicians and/or clinical services.

As friends, employers and other social contacts have greater awareness of the participants' situation, empathy, and the capacity to adjust to their needs accordingly allowed participants to regain their status within these social networks and contributed to helping others who were similarly affected by ICCs. The increased sense of

relatedness fostered perceived competence giving rise to more self-directed behaviours and thus, a greater sense of autonomy (Deci and Ryan, 2014).

A deleterious effect can be seen when there is alienation with the family; lack of clarity, unreliability and mistrust in clinical staff and systems and social pressure that removes a sense of choice and autonomy. However, in this study, most of the participants were able to find alternative sources of support whether these were in place of immediate family and friends; and in the case of clinical staff, usually it was the CGN who was relied on for support.

7.4 Implications for Intervention Design

The application of SDT to this study has highlighted targets for intervention in terms of the basic psychological needs: competence, autonomy, and relatedness. In increasing the satisfaction of these basic needs through health education, providing an autonomy supportive health environment, and developing support networks, this will fulfil the aims of the psychoeducational intervention to support optimal health, informed decision-making, timely adjustment to health status and maximal coping strategies.

This qualitative study builds on the evidence generated by the systematic review in Chapter 3. The findings reinforced the health education components mentioned in the systematic review and added emphasis to tailoring of information. Initial assessments should support the development of a patient profile which will enable consideration of past and current health behaviours and preferences. This would also enable tailoring to age groups as although the intervention is being developed for adults, it is recognised that there may be concerns unique across age groups for example, pregnancy in the 20-30s age bracket. Alongside the direct recommendations from the participants regarding the psychoeducational intervention described in Chapter 6,

these were taken forward in designing the intervention in the Modelling phase of the intervention (Chapter 8).

In terms of outcome measures for the intervention, the application of SDT has paved the way for consideration of outcome measures used in SDT-based interventions. These will also be discussed further in the Modelling phase of the intervention, as part of the synthesis of the evidence base from this study and the systematic review.

7.5 Implications for practice and further research

This study has given an in-depth report on the experiences of family members at-risk for ICCs as they go through cardiac screening and/or PGT in two major established ICC specialist centres. Whilst the infrastructure and personnel for a comprehensive ICC service were in place in these centres, there are still many ways to improve care for this patient group. In accordance with important time periods in the patients' journey, suggestions for improvement in clinical services are summarised in Table 11.

An important point highlighted by this study is the need for clear communication of the purpose of the ICC clinic and to make it explicit to prospective patients that engagement does not equate to undergoing tests but to enable them to make informed decisions about their ICC risk.

The manner of delivering health information and feedback is just as important as the quality of this information. A follow-up phone call or access to a member of the ICC team, particularly when a patient is diagnosed or found to be a genetic carrier, gives the patient a period of reflection and be in a better position to ask questions. In this study setting, it was the CGN who provided much of this support and coordination of care and thus, the need to have this role and resource available more widely was strongly supported by the participants.

A ‘one-stop’ clinic was highly favoured; however, this could be improved by allowing families to attend as a group both for convenience but also to provide support for each other. Most participants felt they received excellent service from the ICC clinic but have had negative experiences with other healthcare providers. Thus, this has made some wary of approaching their GP or other local services. Better coordination, awareness and training and development of networks will be required to ensure ICC care is sustainable and some aspects of ICC care can be provided outside of a specialist clinic.

The development of the psychoeducational intervention through this research will address the need for specific interventions to support the health education and psychological support needs for this patient group. However, the narrative of bereaved families and parents of severely affected probands may need further study to develop strategies to support them in these additional aspects.

Communicating the risk for sudden death is also an important aspect of patient care that has been identified in this study that requires further research not only because this is consistently present in participants’ minds to some degree but also, they have reported that this tends to be downplayed by health professionals especially if the risk is low.

Overall, expertise in the management of ICCs has improved allowing patients and families to have trust and confidence in their care but many improvements in terms of the infrastructure and patient pathways; and communication can be implemented to ensure more equitable, coordinated care. A point of contact for the ICC health care team and source of initial support in the form of the CGN has been recommended as a standard for care. As well as providing further justification to develop the

psychoeducational intervention, this study has also revealed that more research should be undertaken to focus on specific sub-groups within this population, as well as in the communication of sudden cardiac death risk.

Table 11 Recommendations for improving inherited cardiac condition services

Time Period in the ICC Clinical Pathway	Existing good practice or suggestions for improvements
Pre-clinic (initial awareness of risk and referral)	• Emphasis on engagement rather than performance of tests • Comprehensive information regarding clinic procedures, tests, and timelines • Highlight information needed from patients-family history and post-mortem/medical reports
Initial clinic attendance	• Expert medical input • Family attendance but provision for private consultations especially for young adults • One-stop service for tests • Clear and simple, practical instructions regarding clinical tests • Unambiguous feedback and clinical plan after tests
Receipt of results & immediate follow up	• Letter with abnormal/uncertain findings, diagnosis or positive genetic carrier status followed up with a phone call • Point of contact for access to ICC team • Support for cascade screening of relatives • Signposting to patient support groups
Long-term follow up	• Availability of all clinical information at clinic visit • Consultation with named consultant • Point of contact for access to ICC team • Better system of recall for follow-up visits (especially for 3–5-year intervals) • Improved coordination with GP and other local services • Good communication with wider health team-paediatrics, obstetrics, clinical genetics

7.6 Strengths and Limitations

Whilst this study has presented an in-depth account of the experiences of relatives at risk for an ICC as they undergo cardiac screening and/or PGT, it is still important to recognise its limitations. A key aspect to consider in conducting qualitative research is whether the study sample was appropriate in achieving the study aims (Denzin and Lincoln, 2011).

This study incorporated the views and experiences of patients who have undergone screening and/or PGT. It is a known issue in recruitment studies that there is some bias in those who agree to participate. To counteract this, sampling frames were used to encourage sample heterogeneity. This resulted in a population with a representation across genders, age, ICC conditions, cardiac screening and/or PGT processes and results, and symptoms, which are all likely to impact on decision-making. However, there were more participants at-risk for HCM and a preponderance on cardiomyopathies rather than inherited arrhythmias, which is consistent with the higher prevalence of this condition within ICCs. As more patients are identified with other types of ICCs, this may pave the way for better sampling across conditions. Also, there was only one participant recruited who decided not to pursue cardiac screening initially and this is reflective of the finding that in general, there is the belief that attendance in an ICC clinic will automatically result in screening and/or PGT. Despite these limitations, the views collected in this study were wide-ranging whilst generating sufficient data for the identification of consistent reoccurring themes.

The conduct of the group interviews within family groups also had the potential of restricting individual views due to authority roles, particularly amongst parents. As this was acknowledged prior to data collection, interview techniques such as probing and the provision of opportunities and ample time to speak supported the expression of

individual opinions. Furthermore, the family groups had the advantage of bringing forth unique insights on how families support each other.

This study was conducted in two established ICC centres in an urban setting and may therefore limit the applicability to more suburban or rural settings with less developed ICC services. However, the single geographical focus may also be considered an advantage of this research due to the varied backgrounds of those attending these busy clinics as well as identifying issues present in established ICC centres which could be supportive to those in the early stages of developing their ICC services.

Another important aspect that could be a source of bias for this study is the researcher who was known as having the role of a CGN by the participants. Negative views regarding the ICC service and personnel could have been suppressed and overly positive views may have been expressed by participants as they associate the researcher with ICC services. To counteract this, there was a careful distinction of this research from routine care as part of the consenting process and the researcher was not in a clinical role at the time of the study. This did not seem to bring about issues as views expressed by the participants (often quite candid and passionate) regarding their care spanned the breadth of positive and negative descriptors whilst eliciting the quality of relationships they had with their healthcare team.

Researcher bias may have also come about during the collection and interpretation of data. To counteract this, measures implemented included a second researcher reviewing the transcripts, conducting parallel coding, and checking the development of themes through an iterative process. Furthermore, there was support from the supervisory team through all stages of the Framework Analysis approach in terms of coding, mapping, and interpretation. Therefore, adherence to best practice guidelines supported the analysis to ensure good fidelity in relation to the raw data.

7.7 Summary and conclusion

This study was able to provide an in-depth account of the views, experiences, and preferences of relatives at-risk for an ICC undergoing cardiac screening and/or PGT. The main themes generated, Impact of the proband's story, Leveraged autonomy, Harnessing competence and Relatedness in the social context of ICCs detailed their journey to adjusting and coping with their ICC status across key timepoints from the pre-clinic period up to the period of longer-term follow up. The key components of the intervention were also more defined in terms of the health education and psychological support aspects, as well as considerations for format and timing.

The application of SDT as a theoretical framework was also instrumental in explaining the phenomena of how this adjustment and coping developed in individuals and families and therefore, helped identify targets for the psychological intervention as part of fulfilling the basic psychological needs of competence, autonomy, and relatedness.

This study has generated new evidence that will be synthesised with the evidence generated by the systematic review reported in Chapter 3 to develop a psychoeducational intervention model to support at-risk family members as they undergo cardiac screening and/or PGT which will be discussed in Chapter 8.

Chapter 8 Phase 3: Modelling process and outcomes

8.1 Introduction

This chapter presents the synthesis of evidence for the psychoeducational intervention generated from the systematic review and qualitative study, incorporating the application of SDT as a theoretical framework. The logic model for the components and the mechanism of the intervention is presented followed by a report of the consensus exercise with the PPI group. This brought forth the prototype for psychoeducational intervention model with recommended outcomes. This concludes the scope of this research within the MRC Framework.

8.2 Aim and objectives

The aim of this stage was to develop a psychoeducational intervention model.

The specific objectives were to:

- Integrate theoretical findings on the essential components and features for a psychoeducational intervention for at-risk family members as they undergo screening and/or PGT for an ICC
- Incorporate experts-by-experience inputs and end user preferences for the intervention using an iterative consensus approach

8.3 Evidence synthesis from Phase 1 and 2

This research was conducted within the MRC Framework using a mixed method multi-phase approach which allowed each sequential study to build on previous knowledge (Cresswell and Plano Clark, 2017). The systematic review established a preliminary evidence base for the intervention by eliciting the psychological impact and experiences of family members at-risk for ICCs as they undergo cardiac screening and/or PGT. The important role of the family in decision-making, early assessment of

psychological status and needs; and groups that that tend to have increased anxiety and poorer quality of life were highlighted. Key health education components for the intervention, including the need for tailored lifestyle and management advice were also identified. Outcome measurement tools were also examined.

The qualitative study gave greater insight on the contemporary experiences of at-risk relatives as they undergo cardiac screening and/or PGT within a specialist clinic. Apart from reinforcing the findings from the systematic review, additional evidence on the psychological component, format and delivery of the intervention was generated. Greater theoretical understanding of how at-risk relatives cope and adjust to their ICC status was also accomplished through the application of SDT as a theoretical framework. This also paved the way for exploring outcomes specifically designed for SDT interventions.

The synthesis of the findings of the systematic review and the qualitative study was guided by the principles of integration of results in mixed methods studies (Cresswell and Plano Clark, 2017). This is summarised and presented in a joint display in Table 12. The initial step was to search for concepts and themes across the two sets of findings across the key features for the intervention including content, format and delivery, access and duration, and timing. All options for each intervention feature were included for each set of findings.

Within the joint display, the findings were presented to highlight the themes within the systematic review and then sequentially as to what the qualitative study has added to the initial set of findings (Plano Clark et al., 2009). No incongruence was noted in this process. Once the key components and features of the psychoeducational intervention were elicited, the basic psychological needs of competence, autonomy, and relatedness within SDT to which they applied were identified and also noted in

the joint display. An overall recommendation was then concluded for the psychoeducational intervention.

The key components and features identified for the psychoeducational intervention oftentimes overlapped in how they could support the satisfaction of the basic psychological needs. Some examples include health education components that mainly addressed the need for competence, however, a good understanding of ICCs and management options also supports autonomous decision-making. Improving problem-solving strategies also promotes autonomy whilst building confidence and competence. A peer support network not only fosters relatedness but also develops competence as peers learn from each other. Facilitating coping skills as part of psychological support can address all the basic psychological needs as competence arises from seeking knowledge, relatedness develops from building support networks and autonomy is promoted by providing options.

The synthesis of the findings presented in the joint display facilitated the construction of the logic model for the intervention (Figure 11). The logic model maps out the intervention inputs and activities and the proposed links between the intervention and the expected outcomes to summarise a theory of how an intervention might work. The key ingredients for the intervention from the evidence synthesis comprise the inputs for the logic model which in turn defined core activities to undertake for the intervention. These activities were also based on the synthesised findings as well as the guidance for creating a psychological need supportive healthcare environment based on SDT (La Guardia, 2017).

The logic model proposes that by providing these SDT-based components and actions within the social context of the individual, they will have increased knowledge, stronger family and social networks, greater sense of confidence and choice, which

will in turn, increase the satisfaction of and result in better outcomes for the basic psychological needs of autonomy, competence, and relatedness. Overall, the at-risk family member undergoing cardiac screening and/or PGT for an ICC will have a greater degree of self-determination, decreased anxiety and be able to adjust and cope with their situation more optimally.

Table 12 Synthesis of Systematic review and Qualitative study findings

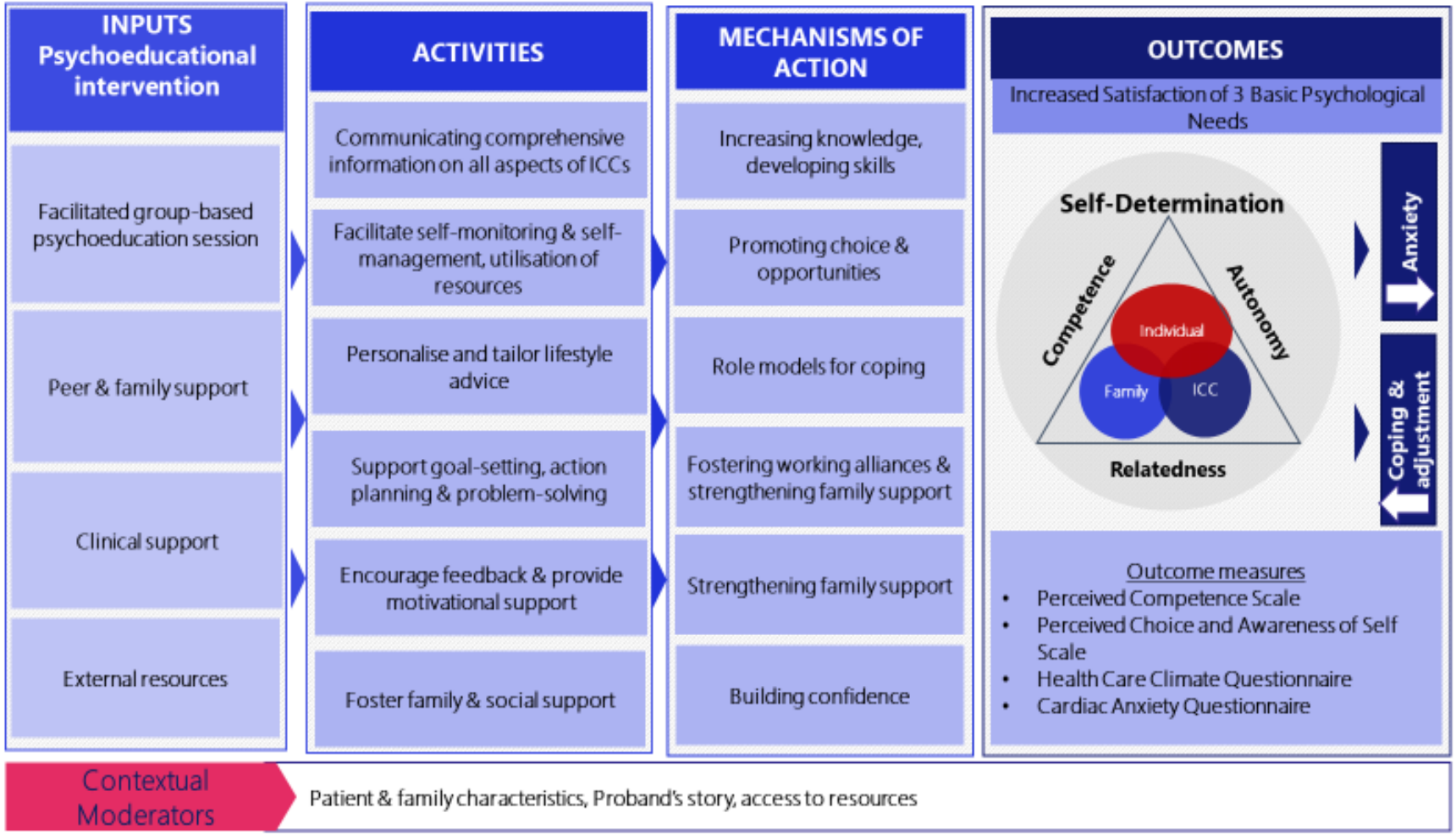
Key content and implementation strategies	Research Evidence		Overall recommendation for the psychoeducational intervention
	Systematic review	**Qualitative study**	
Content	➢ General inherited cardiac conditions (ICC) knowledge, including genetics **(Competence-C)** ➢ Key areas highlighted for health education: • Symptom identification & management **(C)** • Risk for sudden cardiac death **(C)** • Medication & devices **(C)** • Tailored advice according to ICC status, interests, social practices and age **(Autonomy-A, C)** ➢ Psychological component • Autonomy support **(A)** • Problem solving strategies **(A, C)** • Coping skills **(A, C, Relatedness-R)**	➢ Consistent with systematic review for health education content as participants were also at-risk family members in the included studies with the addition of information regarding ICC care pathway and access ➢ Added components identified for supporting psychological well-being • Communication skills **(A, C, R)** • Building confidence **(A, C)** • Peer support **(R)**	➢ Comprehensive combined psychoeducation content with flexibility for tailored advice **(A, C, R)**

Key content and implementation strategies	Research Evidence		Overall recommendation for the psychoeducational intervention
	Systematic review	**Qualitative study**	
Format and delivery mode	➢ Peer support or signposting to patient support groups **(C, R)**	➢ Leaflets or online format of educational materials **(C)** ➢ One-to-one support from the healthcare team in person, by telephone email, likely to be the cardiac genetic nurse (CGN) **(A, C, R)** ➢ Facilitated group format mixed with peer support **(C, R)** ➢ Delivered within the family as a group or groups of families **(C, R)**	➢ Use of expertly curated printed and online resources already widely available **(C)** ➢ Peer and/or family group format facilitated by the CGN with access to ad hoc telephone or email support. **(A, C, R)**

Key content and implementation strategies	Research Evidence		Overall recommendation for the psychoeducational intervention
	Systematic review	**Qualitative study**	
Access and duration	Not noted	➢ Offered as part of the care pathway either as a programme or ad hoc support line **(A, C)** ➢ Available throughout long-term follow up **(C, R)**	➢ Positioned as part of the ICC care pathway with telephone helpline available throughout follow up **(R)**
Timing	➢ Assessment of needs psychological status and needs as early as possible **(A, C)** ➢ Likely to be needed when waiting for or when results are received **(C)**	➢ As early as possible but emphasis on the immediate period after a diagnosis and/or predictive genetic test (PGT) results **(A, C)**	➢ Early assessment of psychological status and needs **(A, C, R)** ➢ Main group session timed immediately after diagnosis and/or PGT results **(C, R)**

Key content and implementation strategies	Research Evidence		Overall recommendation for the psychoeducational intervention
	Systematic review	**Qualitative study**	
Implementation considerations	➢ Consideration for needs of certain groups: Bereaved families and parents **(A, C)** ➢ In non-UK settings, may have insurance implications **(A, C)**	➢ Facilitated as a group but with flexibility for individual approach **(A, C, R)** ➢ Same groups identified as the systematic review for those who might have additional needs ➢ Possible dominance of parents in family groups **(A)**	➢ Expert facilitation **(A, C)** ➢ Ground rules to ensure confidentiality and non-pressurised atmosphere **(C, R)**

Figure 11 Logic model for the psychoeducational intervention



8.4 Patient and public involvement activities for the intervention model

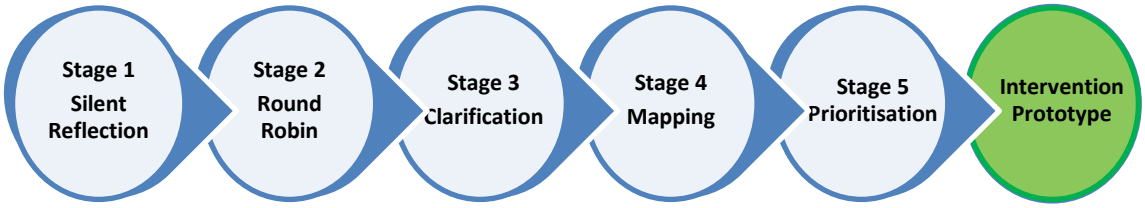
The KHP-PPIICC group provided input all throughout the planning and conduct of the research. They played a major part in developing the proposed intervention within the final, modelling phase of this project as part of the Development stage of the MRC Complex Intervention Framework. The group is comprised of ICC patients, family members and ICC clinicians who have experience of the cardiac screening and/or PGT process. Details on the establishment and composition of the group was described in Section 2.3.2.

In the development of the intervention model, the group undertook a consensus exercise to assist in the mapping and prioritisation of the components, delivery methods and other aspects identified in the synthesis of evidence from the theoretical work done thus far. This ensured that the intervention would be relevant, useful, and appropriate for those who would be delivering and receiving the intervention.

8.4.1 Modified Nominal group technique

A modified nominal group technique (NGT) adapted from Perry and Linsley (2006) was used by the KHP-PPIICC group to arrive at a consensus regarding the components and features of the intervention model. The NGT is an evaluative methodology wherein participant involvement is non-hierarchical and responses are given equal validity. The approach consisted of several stages as summarised in Figure 12. The NGT is part of the PPI activities.

Figure 12 Stages of the Nominal Group Technique



The main NGT session was held on the 16th of July 2018 and attended by ten members of the KHP-PPIICC group, including one CGN. A further three members who could not attend in person provided feedback via email. At this point, the group was already familiar with the study having had briefings since June 2017.

The session started with the Silent Reflection stage where the group was presented with a preliminary matrix of the evidence synthesis, logic model and outcome measures from the previous theoretical work done and the more specific suggestions from the participants in the qualitative study. They were also shown examples of ICC information leaflets from the BHF (Appendix 7 A) as a form of readily available patient resources and a personalised lifestyle form modified from the European Society for Cardiology (ESC) Guidelines for the diagnosis and management of HCM (Elliott et al., 2014) that can be used to tailor lifestyle advice (Appendix 7 B). They were asked to consider the components, content, and other features that they felt would be relevant to the intervention and would be helpful to patients undergoing cardiac screening and/or PGT.

In the Round Robin stage (Stage 2), the group was asked to write as many components or features for the intervention that they believed to be pertinent based on their reflections on individual sticky notes. These were then posted on a flip chart. The Clarification stage (Stage 3) followed where the researcher approached each group member for any additions and to clarify any terms on the notes that were unclear.

The notes were then mapped (Stage 4) according to the components and features of the intervention such as content, delivery, timing and so on. As the mapping progressed, discussions led into Stage 5 where prioritisation of elements within components and features were done simultaneously, particularly if it was obvious from the note count and responses from group that these were essential.

8.4.1.1 Components and features of the intervention

The main responses from the group were focused on the content of the intervention. It was felt that the BHF leaflets were adequate as it covered all the basic information on the ICCs from genetics, pathophysiology, diagnosis, and management. These topics were all deemed essential by the group members. The BHF leaflets were developed with clinicians and patients, updated regularly and were freely available as hard copies and online. Therefore, these would be adequate in helping to provide basic information to patients. What the group felt was important was the tailoring of advice for individuals which the form adapted from the ESC HCM guidelines could facilitate as this could be used as prompts to reflect on their current situation and goals and actions for any adjustments that needed to be made. Of all the features of the questionnaire, medications and side effects were felt to be the most important to the group.

Other elements for the content of the intervention that were thought to be a priority were the familiarisation with the ICC service and process of long-term follow up. This harked to the need for relatedness to grow between patient and clinician. The group also felt that support for communication of risk with family members should also be part of the intervention as this is not always easy and straightforward. Links and information about support groups in the community should also be part of essential information for patients.

In terms of timing and delivery, the group felt that those who would benefit from the intervention were those who were newly diagnosed or found to be carriers for an ICC and so should be implemented not long after the patient receives their test results. A consensus for a group approach for the intervention, with facilitation from an experienced CGN, was reached as it was felt that this would help patients support each other but also to provide a structure within which patients will feel safe and not be forced to share anything they would not want to. The group was unsure on the number of sessions that should be offered but considering that most patients may be working or must travel if the session was held in the hospital, it was agreed that one 2-hour session would be practicable. Both patient and clinician members of the group felt that it was a practical and efficient way to deliver the intervention to more patients, but also agreed that the group size should not be too big, at most 12 attendees at a time and that the existing CGN helpline that is part of the clinical service should continue to be in place. The clinicians in the group also felt that this was feasible in practice within their ICC service and would be an opportunity to have more insight into how their patients are coping with the ICCs.

In terms of group composition for the intervention, apart from the patient, it was felt that a family member or significant other could also attend either to listen or participate directly as this will also support their role and competence in the context of the ICC in the family and build relatedness.

Whilst it was felt that patients with different types of ICCs could be mixed in the groups, one young member of the group felt that sessions by age group would enable more tailoring to their needs for example childbearing, career, and exercise. Overall, whilst the group agreed that there are topics that would be more pertinent to young people, they did not feel this was the right approach as both young and old can learn

from each other in terms of their experiences, and in particular how the older patients have coped in their youth (though they may not have been aware that they had an ICC at that time).

Other aspects of the delivery of the intervention that were discussed included types of activities that could be done. This included a question-and-answer session mostly for medical concerns, role playing or workshops that could facilitate skills and confidence when speaking to clinicians and family members, and a patient speaker who can talk about their patient journey after a diagnosis or carrier status for an ICC.

Finally, the group commented on the outcome measures for the intervention that were selected based on the evidence synthesis from the systematic review and the qualitative study.

8.4.1.2 Outcome measures

8.4.1.2.1 Outcomes based on Self-Determination Theory

Self-Determination Theory has been used throughout as the theoretical framework for the psychoeducational intervention. It has been postulated that the satisfaction of the three basic psychological needs of competence, relatedness and autonomy will bring about decreased anxiety and increased coping and adjustment to the ICC diagnosis and/or carrier status. There are many validated outcome measures within SDT in the field of healthcare to elicit the degree of satisfaction of the basic psychological needs, however, none have been directly applied to ICCs. It was therefore, felt necessary to consult with experts in SDT to adapt the questionnaires to ensure applicability to the ICC patient group. This consultation was undertaken with two SDT experts from the School of Health and Rehabilitation Sciences, University of

Queensland in Brisbane, Australia, Professor Jenny Ziviani and Professor Anne Poulsen.

Perceived Competence Scale

The Perceived Competence Scale (PCS) is a short, 4-item questionnaire, and is one of the strongest for face validity amongst the instruments designed to assess constructs from SDT (Smith et al., 1995). The PCS assesses participants' feelings of competence about, for example, taking a particular college course, engaging in a healthier behaviour, participating in a physical activity regularly, or following through on some commitment. The PCS is typically written to be specific to the relevant behaviour or domain being studied and as such, this was modified to focus on ICC health management (Appendix 8 A) in consultation with Professor Ziviani and Professor Poulsen. For this study, the questionnaire was modified to reflect the feelings of competence for coping with the management regime and challenges of the ICC diagnosis or genetic carrier status.

Perceived Choice and Awareness of Self Scale

The Perceived Choice and Awareness of Self Scale (PCASS) (Appendix 8 B) looks at individual differences (trait level) in perceived choice and awareness of self (Silva et al., 2010). The PCASS is a short, 10-item scale, with two 5-item subscales. The first subscale is perceived choice in one's actions, and the second is awareness of oneself. The subscales can either be used separately or they can be combined into an overall score. Perceived choice reflects feeling a sense of autonomy and choice with respect to one's behaviour and awareness of self reflects being aware of one's feelings and sense of self.

Health Care Climate Questionnaire

The Health Care Climate Questionnaire (HCCQ) has a long form containing 15 items and a short form containing 6 of the items. Specifically, depending on the issue being examined, the HCCQ can be used to assess relatedness and the patients' perceptions of the degree to which their specific doctor is autonomy supportive, or it can be used to assess patients' perceptions of the degree to which their team of health care providers is autonomy supportive (Czajkowska et al., 2017). The latter is typically used if patients are seeing several providers within a clinic to deal with a particular problem such as the multidisciplinary team in a typical ICC clinic. However, the relationship with the cardiologist and the CGN was most frequently mentioned in the patient interviews and therefore it was felt that it was important to ascertain the relation between these two types of healthcare providers' interpersonal style and their patients' motivation, behaviour, and health. Professor Ziviani and Professor Paulsen supported the use of the 15-item version of the questionnaire, worded in terms of 'my doctor 'and my 'cardiac genetic nurse' (Appendix 8 C) as this questionnaire is typically modified in this manner depending on the healthcare practitioner.

8.4.2.1.2 Cardiac Anxiety Questionnaire

The Cardiac Anxiety Questionnaire (CAQ) (Appendix 8 D) is an 18-item, self-reported questionnaire, designed to measure heart focused anxiety, rated on a 5-point Likert scale ranging from 0 (never) to 4 (always) (Eifert et al., 2000). Heart-focused anxiety (HFA) was defined as "the fear of cardiac-related stimuli and sensations because of their perceived negative consequences". Heart-focused anxiety may apply to medical conditions characterised by chest pain and psychological distress, among which are cardiac and non-cardiac chest pain and panic disorder. Eifert et al. (2000) have developed the CAQ to specifically assess cardiac anxiety, heart-focused attention, and related avoidance behaviours. They showed that the CAQ to have adequate

psychometric properties and was able to differentiate cardiac from general anxiety in psychiatric outpatients.

The CAQ has been used in a prospective study on patients with LQTS and HCM and found that those with a diagnosis have higher scores than those who were asymptomatic genetic carriers in the three distinct aspects of HFA-avoidance, attention, and fear. As well as helping to identify patients who may be unnecessarily avoiding activities that could be health promoting, using the CAQ as an outcome for the psychoeducational intervention will help assess effectivity in alleviating stress and anxiety.

8.4.2.1.3 Patient and public involvement group feedback on the outcome questionnaires

Overall, the proposed outcome questionnaires were described as appropriate and not too taxing by the PPI group. The ICC patients in the PPI group felt that there will be points where patients might not be keen to answer the questionnaires (periods of denial, frustration, or being unwell). Therefore, adequate time should be given for the return of questionnaires balanced with the need to capture the patient's status at the timepoint being captured by the questionnaire.

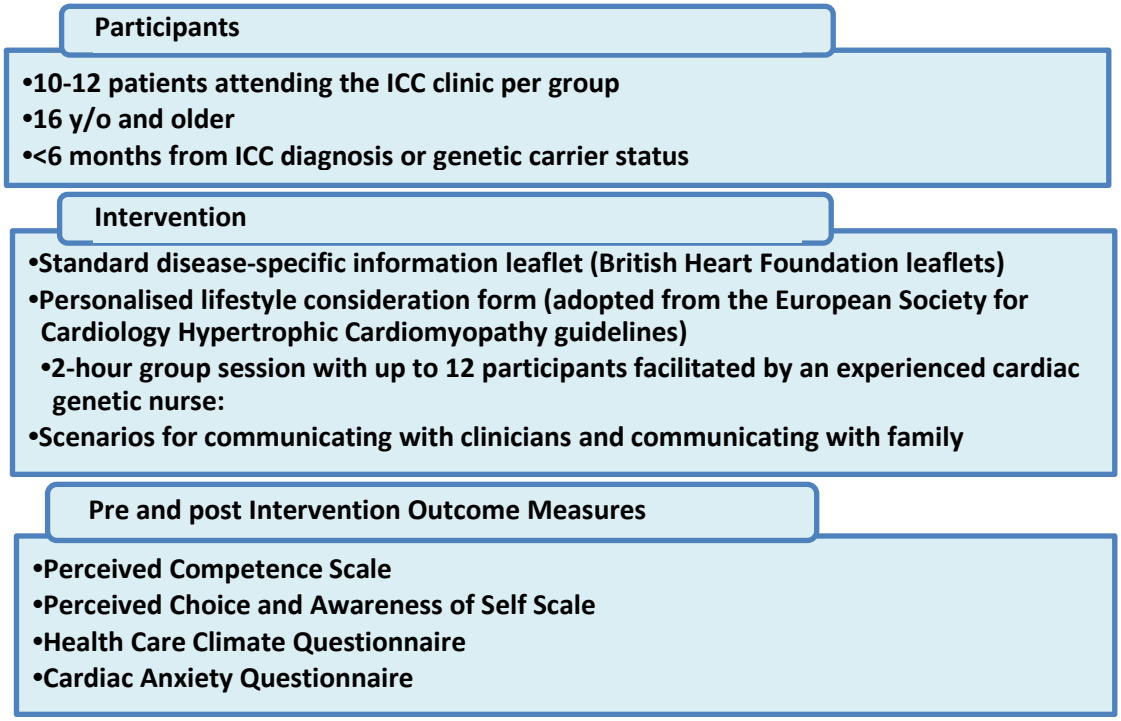
The PPI group felt that the SDT-based questionnaires reflected the basic psychological needs satisfaction but that support for the context of the questionnaire should be explained. For example, for the PCS, it should be made clear that there is no expected level of competence at a particular timepoint. The patients and relatives amongst the group also emphasised that the HCCQ was important because developing a good working relationship with their clinicians was key to their own and their children's coping.

The CAQ also stood out for the group as something that would help patients reflect on how their cardiac-associated symptoms are impacting on their daily life as well as measure any improvements that may be due to the intervention over time.

8.5 Intervention model prototype

The consensus exercise conducted with the PPI group based on the synthesised evidence from the systematic review and the qualitative study has enabled the proposal of a psychoeducational intervention model prototype. In this section, the content and features of the prototype is discussed and is summarised in Figure 13.

Figure 13 Psychoeducational Intervention Supporting Patients with Inherited Cardiac Conditions (PISICC) intervention prototype



8.5.1 Participants and timing of intervention

Overall, based on the synthesised evidence and the PPI group opinion, the target population for the psychoeducational intervention should be focused on patients who have a new diagnosis and/or carrier status for an ICC. This will enable early

assessments and provision of tailored health information and support at a time when most patients tend to be most anxious. In addition, this helps foster effective communication with the healthcare team and their families at a period when many decisions are made regarding the next steps for their health and further family screening. Family members may be invited to the sessions in a supportive role for the patient.

8.5.2 Content and delivery

The content for the psychoeducational intervention consists of two parts: an information and health education component and a component to support psychological well-being. In combination, these support the fulfilment of the basic psychological needs of autonomy, competence, and relatedness.

Information about ICCs is widely available on various websites and can be ordered in print from charities like the BHF and other patient support groups. The disease specific information leaflets provided by the BHF is regularly updated and provides reliable general information on the ICCs in terms of pathophysiology, genetics, and medical management.

Tailoring of information requires development of a patient profile which will enable consideration of past and current health behaviours and preferences. This would enable tailoring to age groups as it is recognised that there may be concerns unique needs across age groups for example, pregnancy in the 20-30s age bracket. The personalised lifestyle consideration form (based on the ESC HCM guidelines) and the question-and-answer session, enables the provision of tailored health information by an experienced CGN according to the participants' needs, goals and preferences in terms of lifestyle adjustments, medications, and symptom management.

To support psychological well-being, the content of the intervention should incorporate strategies that enhance the achievement of tasks that have been identified as key to coping such as the ability to communicate effectively with clinicians about their care; communication of risk information with other family members; acceptance of their ICC status and actions taken to achieve a sense of normality. The group format wherein these scenarios are discussed encourages peer support and helps bring forth many options on how to communicate effectively.

The intervention should continue alongside the routine care provided by the CGN which typically consists of a telephone helpline, availability of ad hoc consultations and signposting to patient support groups.

8.5.3 Outcome measures

The outcome measures discussed in Section 8.4.1.2 based on SDT (Perceived Competence Scale, Perceived Choice and Awareness of Self Scale, Health Care Climate Questionnaire) would evaluate the efficacy of the intervention and identify areas of the basic psychological needs that need further support. The Cardiac anxiety questionnaire can also assist in instigating investigations for ongoing physical symptoms and referrals to other services such as clinical psychology should levels remain elevated.

8.6 Summary and conclusion for Phase 3 Modelling process and outcomes

The modelling process and outcomes phase is the final step covered within the Development Stage of the MRC Framework for the design and evaluation of complex interventions within this project. By completing this phase, the PISICC project has progressed from establishing the evidence and developing theory to the development of a psychoeducational model prototype. During this cross-phase work, patients and

family members with lived experiences of ICCs, as well as healthcare professionals directly involved in ICCs, were engaged through a consultative and consensus process to bring about an intervention model and potential outcome measures that are likely to be acceptable to patients and ready for feasibility testing.

The iterative process in the three phases of the PISICC project sets out clearly the indicative content and features for the intervention, however, to determine effectiveness, a clinical trial would be the gold standard to support adoption of the intervention in routine clinical practice. Prior to this major undertaking the MRC Framework calls for the fulfilment of further stages of intervention development and thus feasibility and/or piloting studies for the intervention would be the next steps to progress the PISICC study.

Chapter 9: Discussion & conclusion for the PISICC project

9.1 Introduction to the final chapter

Chapter 9 draws together the findings from the PISICC project, in particular, findings from the qualitative project and discusses them in the context of updated reviews of similar research. The results of the various phases of the PISICC project are summarised in Section 9.2. In Section 9.3 the key findings are discussed in relation to the research objectives and wider literature followed by Section 9.4 wherein the project is critiqued to identify strengths and limitations. The implications and new knowledge generated by the research is then discussed leading to recommendations for clinical practice (Section 9.5) and further research in the area (Section 9.6). The final the conclusions for the project are presented in Section 9.7. and dissemination and awards thus far are listed in Section 9.8.

9.2 Summary of main findings

This thesis reports on the development of a psychoeducational intervention to support at-risk family members undergoing screening and/or predictive genetic testing for an inherited cardiac condition, using a mixed methods approach within the methodological framework of the MRC Framework for Developing and Evaluating Complex Interventions (Craig et al., 2008). The project incorporated three distinct but inter-related phases with a concomitant feedback loop for theory development. These stages corresponded to the Development stage of the MRC framework addressing the main aims of developing a healthcare intervention incorporating patient education and psychological support optimised according to the preferences of end-users.

Having set out the background to the study and summarised the need for the research in this area in Chapters 1 and 2, Phase 1 of the PISICC project consisted of a systematic review (Chapter 3) which established the preliminary evidence base for

developing the psychoeducational intervention. The synthesis of evidence from both quantitative and qualitative primary studies highlighted the experiences and gaps in service provision for at-risk patients undergoing screening and/or PGT for ICCs brought forth possible components for an intervention.

Recommendations for the psychological component included early assessment of psychological status and needs. The exploration of perceptions, values and preferences, autonomy support; and harnessing support from family or social groups are also key ingredients. For the health education component, information about the management of signs and symptoms, indications for medications/devices and dealing with side effects should be incorporated. Possible outcome measures were also identified to measure effectiveness and acceptability of an intervention.

The core themes generated from the systematic review: Family, Psychosocial adjustment and Autonomy, led to the exploration of Self-determination theory (Ryan and Deci, 2017) as a theoretical framework from which to develop the intervention. Improving self-efficacy is built into the concept of Competence as this encompasses the health information and coping skills that individuals need to have confidence in managing their health. The importance family relationships and shared experiences alongside the interactions with health professionals and other social connections are captured in Relatedness. The need to support intrinsic motivation in decision making across all aspects of ICCs is emphasised in Autonomy.

Alongside functioning as a framework to identify components and targets for the psychoeducational intervention, SDT was also proposed to explain the phenomenon of adjustment and adaptation that occurs in at-risk ICC patients who are diagnosed with an ICC and/or found to be genetic carriers. This is due to SDT's assumption that satisfaction for the basic psychological needs of competence, autonomy and

relatedness is the essence of human thriving and contribute to indicators of wellness and vitality (Ryan and Deci, 2000).

Determining the theoretical framework for the intervention occurred concurrently along the three phases of this project as each piece of work helped to provide a feedback loop. This ensured that SDT was consistent with the findings from each phase and served as an important check at each step as this was a novel application to genetic healthcare.

The systematic review emphasised health advice and psychological assessment and support as components for an intervention, however, the form and manner by which psychoeducational interventions should be delivered was not elicited. In Phase 2, a qualitative study (Chapters 5-7) was undertaken to build on this evidence focused on the experiences of at-risk family members who have undergone screening and/or PGT for an ICC within a contemporary multi-disciplinary specialist clinic.

The qualitative study was able to provide an in-depth account of the views, experiences, and preferences of this patient group. The main themes generated, Impact of the proband's story, Leveraged autonomy, Harnessing competence and Relatedness in the social context of ICCs detailed their journey to adjusting and coping with their ICC status across key timepoints from the pre-clinic period up to the period of longer-term follow up.

Furthermore, the qualitative study provided more insight into possible modes of delivery with expertly facilitated group sessions acceptable to most participants with the added benefit of learning from their peer group. There was also an overwhelming need for support at the time of diagnosis or receipt of results which can be in the form of a formal programme offered routinely to minimise stigma associated with being

deemed to need ‘psychological support’. Participants also made direct recommendations on how clinical services could be improved.

The application of SDT as the conceptual framework in the analysis of the qualitative study has highlighted targets for intervention in terms of the basic psychological needs: competence, autonomy, and relatedness. In increasing the satisfaction of these basic needs through health education, providing an autonomy supportive health environment, and developing support networks, this will fulfil the aims of the psychoeducational intervention to support optimal health, informed decision-making, timely adjustment to health status and maximal coping strategies. The application of SDT also paved the way for consideration of outcome measures used in SDT-based interventions.

In Phase 3 of this project, the modelling process (Chapter 8) within the MRC framework saw the evidence from systematic review and qualitative study synthesised to develop a logic model for the psychological intervention that is underpinned by SDT. Components and activities for the intervention were designed to support the mechanism of satisfying the basic psychological needs to support coping and adjustment to a new diagnosis or genetic carrier status for an ICC. A formal consensus exercise through the PPI group brought forth the prototype for the psychoeducational intervention model with corresponding outcome measures that can be subjected to the feasibility/piloting phase of the MRC framework.

9.3 Key findings in relation to the research objectives and wider literature

This study set out to develop a healthcare intervention which incorporates patient education and psychological support to address the needs of patients undergoing

cardiac screening and/or PGT for an ICC. To achieve this aim, several objectives were set and were achieved through the three phases of this project.

9.3.1 To establish the evidence base for the psychoeducational intervention

The objective was fulfilled by the synthesis of the evidence from the systematic review and the qualitative study. This fulfilled the first step of the Development stage of the MRC Framework. Findings from the systematic review informed decision-making on the areas of need and components of the intervention whilst findings from the qualitative study gave insight on the timing and method of delivery as well as targets for the intervention and its components.

The systematic review reported in Chapter 3 revealed that there were very few studies that focused solely on the experiences of family members at-risk for an ICC as they undergo cardiac screening and/or PGT as this population was studied alongside probands. An updated literature search has identified two studies where the participants were exclusively family members at-risk for an ICC (Bonner et al., 2018, Bordet et al., 2020) and one study which included both probands and at-risk relatives (Wynn et al., 2018). The study done by Bordet et al. (2020) in France is the largest longitudinal quantitative study so far which comprised of 517 participants divided into a prospective and retrospective cohort. The study conducted by Bonner et al. (2018) is also the largest qualitative study (N=32) thus far focused on the experiences of family members at risk for HCM. Both studies reinforced the findings of this research in terms of concern for children as a main motivator to pursue PGT (above medical concerns) and the lack of children made the need for PGT less urgent.

Similar to the findings in this research, recent studies have shown that there was a very low rate of regret for undergoing PGT and this was felt to be associated with having been done within a specialised ICC service (Bonner et al., 2018, Bordet et al.,

2020). Whilst the findings from Wynn et al. (2018) are consistent with the systematic review and the qualitative study in that those who are found to be genetic carriers for an ICC tend to have higher rates of distress than non-carriers, Bordet et al.'s (2020) study showed that both groups could display high rates of anxiety, and this was correlated to their degree of anxiety at baseline rather than their genetic testing results.

Psychological (shock, worry and uncertainty) and behavioural (career, sport, and insurance) consequences of the cardiac screening/PGT process were still elicited by the current studies (Bonner et al., 2018, Bordet et al., 2020, Wynn et al., 2018) and an emphasis on providing clarity of the clinical consequences of a positive genetic carrier result and informed decision making (Bonner et al., 2018) with specialist teams (Bordet et al., 2020) were reported as key to mitigating the negative impacts .

A key finding in the qualitative study was the significance of a sudden cardiac death in the family in the decision-making for pursuing cardiac screening and/or PGT and that bereaved at-risk family members, particularly parents, are a group requiring specific input. Three cross-sectional survey studies that focus on the impact of sudden cardiac death on at-risk family members add to this evidence, however, it is uncertain whether the participants in these studies overlap as they have recruited from the same ICC registry in Australia (Bates et al., 2019, Ingles et al., 2016, McDonald et al., 2020). In the study by Ingles et al. (2016) scores for severe depression, anxiety and stress were higher in family members who have a relative who died suddenly due to a possible ICC (N=103) compared to the general population. They also found prolonged grief and post-traumatic stress were more prevalent in 19% and 44%, respectively and that this was more common in mothers and those who witnessed the death. This was reinforced by the findings in Bates et al.'s (2019) study wherein decreased well-being, presence of post-traumatic stress and depression were correlated with a

perception of a lack of social and medical support leading to poor adaptation to the results of their deceased relative's molecular autopsy report. McDonald et al.'s (2020) study on 38 parents looked at the unmet needs of this population and medical needs were identified as a foremost priority, however it was the areas of psychological information and support needs that were endorsed by the participants as the most unmet.

The findings of these recent studies support the evidence base generated by this research and provides further justification for the intervention model and the areas of need it is targeting. However, in light of the finding that psychological status pre-PGT is more likely to predict psychological status post result rather than the PGT result itself, it would be important to consider pre-PGT psychological assessment. The additional evidence in terms of the impact of sudden cardiac death and bereavement and the unmet needs in these circumstances brings to the forefront that this group will also need specific support and interventions.

At the time of writing, there were no studies published on the development of a psychoeducational intervention for the at-risk for ICC group. However, a protocol for an RCT to provide a tailored approach to discussing ICC risk with relatives by probands has been proposed (van den Heuvel et al., 2019). The findings of this may be informative in the future as at-risk relatives themselves will need to have this discussion with their own family if they are diagnosed or found to be carriers and this was identified as an important aspect of coping with the results of their cardiac screening/PGT in this research.

9.3.2 To establish the theoretical basis of a psychoeducational intervention within the context of genetic healthcare

The development of the theoretical basis for the psychoeducational intervention was an iterative approach which developed from the needs of family members at-risk for ICCs identified in the systematic review. These needs coincided with the three basic psychological needs-autonomy, competence, and relatedness-the fulfilment of which are central principles in Self-determination theory. The justification for SDT as the theoretical basis of the intervention was made in Chapter 4 and its applicability to the area of ICCs was reinforced as themes emerging from the qualitative study remained consistent with SDT. When used as the conceptual framework in the analysis of the qualitative data, SDT shed light on the phenomena of the adjustment to the impact of ICCs wherein the theme Impact of the proband's story provided a starting point for the participants' narrative and the other themes: Leveraged autonomy, Harnessing competence, Relatedness in the social context of ICCs interact to help fulfil one another to support the individual to grow, adjust and thrive within the ICC landscape.

Whilst the MRC framework stipulates the identification or development of theory to drive intervention development, it does not define a specific method of doing so. The use of a feedback loop to SDT all throughout the phases of this research ensured constant checking for consistency and applicability of the theory for the intervention and helped produce the logic model from which the intervention prototype was derived. This method supports the fulfilment of the application of appropriate theory within the Development stage of the MRC framework.

The novel application of SDT to genetic healthcare expands the scope of this theory and what remains is to test the theoretical assumptions in the logic model through a randomised control trial of the intervention which is outside the scope of this research.

Recent conceptual work on the phenomena of adjustment to an ICC diagnosis includes the application of the Common Sense Model of Illness which postulated that patients are not passive observers in medical consultations; rather, they are constantly seeking, receiving and processing illness-related information from multiple sources (O'Donovan et al., 2020).

For family members who are at-risk for an ICC, there is an emphasis on the dilemma of a positive genotype and no detected phenotype following PGT. The terms 'at-risk relative' or 'asymptomatic carrier' may be experienced as new and perplexing as the absence of clinical signs of the condition is not a typical illness model people can readily make sense of. Perceptions of illness identity is central to patients' conceptualisation of their illness therefore it is easy to see that patients with a genetic diagnosis but no clinical signs of disease may struggle to comprehend and adapt to their condition (O'Donovan et al., 2018). This is an important concept to consider in intervention development as adherence to medical management and lifestyle recommendations may be compromised in a completely asymptomatic ICC genetic carrier. However, in common with Self-efficacy theory (Bandura, 1997) , the degree of relatedness, which is an important aspect in genetic healthcare, and how this influences motivation is not clearly accounted for in this theory.

9.3.3 To determine the components and features essential to the intervention model

The synthesis of the evidence from the systematic review and the qualitative study alongside the development of theory brought forth a logic model and a psychoeducational intervention prototype which satisfied this objective. This iterative method identified the essential components and features essential to the intervention to help support the satisfaction of the basic psychological needs of autonomy,

competence and relatedness to generate the highest form of intrinsic motivation and promote coping and adjustment.

The health education aspects of the intervention, which included information on ICCs, genetics, medical management, and lifestyle recommendations are complemented by the psychological components which involves tailoring of information according to clinical situations and personal goals to increase competence and sense of autonomy. The provision of a group setting provides the opportunity for peer support and the discussion of scenarios identified in this research that participants found most important to build relatedness and competence-conversations with healthcare professionals and with their families.

A systematic review of psychoeducational interventions to support family communication of genetic testing results and cascade screening in hereditary breast cancer/ovarian cancer or Lynch Syndrome showed that many interventions included booklets and family-based communication training but the overall effect size was small and non-significant for the outcome of increased cascade screening/testing (Baroutsou et al., 2021). Therefore, whilst the components for supporting cascade screening are essential the best approach to enhance this outcome is still unknown. It was emphasised that future research should include not just the giver of information (probands) but also recipients of this information. An integrative review of family communication in patients with ICCs also had the same conclusion regarding the inclusion of experiences of at-risk family members on receiving this information (Shah and Daack-Hirsch, 2018). As the population of the PISICC intervention development were at-risk relatives who would have been the recipients of this information, the findings of this research in terms of their experiences upon receiving this information contributes evidence to this research gap.

Furthermore, Baroutsou et al. (2021) noted the reliance on intensive face to face input by a genetic counsellor or health professional for these cascade testing interventions and suggested that in view of the critical shortage of the genetics workforce and the availability of new technologies, web-based interventions should be explored. With the restrictions brought upon the health service by the Coronavirus-19 pandemic, many routine services, including genetic counselling and nursing care were delivered online and will likely be adopted into care beyond the pandemic (Nuthoo, 2020). It may therefore be necessary to explore options for the delivery of the PISICC intervention on an online platform.

9.3.4 To identify outcomes associated with the intervention

This objective was fulfilled through the iterative process in developing and applying SDT as the theoretical framework for the PISICC intervention and through scoping various outcome measures used in the studies included in the systematic review. As the PISICC intervention is aimed at increasing the fulfilment of the three basic psychological needs, existing validated outcome questionnaires used in SDT healthcare interventions were proposed (Perceived Competence Scale, Perceived Choice and Awareness of Self Scale, Health Care Climate Questionnaire). The Cardiac anxiety questionnaire (Eifert et al., 2000) was also proposed as uncertainty around the cause of cardiac-sounding symptoms was cited as a major cause of anxiety and a factor for adjustment in the qualitative study and was related to family history of sudden death and family dynamics in the systematic review. These were deemed suitable and appropriate based on the PPI group activities in Chapter 8 but will require more rigorous testing in an experimental setting as the SDT outcome measures have not been used in this context.

Genetic healthcare-based psychological measures have been recently cited in related cross-sectional studies in family members at risk for ICCs. This included the

Psychological Adaptation to Genetic Information Scale (Bates et al., 2019) and the Multidimensional Impact of Cancer Risk Assessment which was adapted for patients having genetic testing for cardiomyopathies (Wynn et al., 2018). Both these measures seek to determine understanding and impact of receiving genetic information and therefore could be applicable to the PISICC intervention in the areas of adjustment and coping with a genetic diagnosis.

9.3.5 To incorporate the perspectives and preferences of the end-users in the intervention model

Patients and the public have been involved from the inception of this research project. The KHP-PPIICC group is comprised of ICC probands, at risk family members who have undergone cardiac screening and/or PGT and clinicians involved in the ICC service thereby representing both the recipients and the providers of the intervention. Specifically, the KHP-PPIICC group have been instrumental in ensuring the readability of recruitment materials for the qualitative study as well as providing input to the topic guide for the individual interviews and group discussions. This has supported participant recruitment to target as well as a broad dataset from which to draw evidence for the intervention.

The perspectives and preferences of end-users for the intervention were directly captured in the qualitative study in three ways: through a sample of participants who are themselves at-risk family members and have gone through cardiac screening and/or PGT, generation of themes from the qualitative data which identified targets and possible mechanisms for the intervention; and the direct recommendations of the participants on how a psychoeducational intervention might look like and its implementation.

Furthermore, the KHP-PPIICC group also played a key role in eliciting the intervention prototype following the synthesis of evidence from systematic review and the qualitative study through a modified consensus approach described in Chapter 8. This process allowed for the prioritisation of components of the intervention, and the practicalities of delivering it both from the point of view of patients and clinicians. Outcome measures were also reviewed by KHP-PPIICC group for applicability and recommendations on how these should be presented to participants to ensure that there is no expected level/status for any of the outcome scales. A feasibility study for the intervention could provide further input from participants and clinicians in terms of applicability and acceptability to end-users according to the MRC framework but this is beyond the scope of this research.

Patient and public involvement is now recognised as an integral component of healthcare research to ensure the quality and relevance of the research. An example are the James Lind Alliance Priority Setting Partnerships (JLA PSP) which are conducted with a broad range of stakeholders (including patients and the public) to identify uncertainties in evidence which can be addressed by research in a particular area or disease spectrum (Viergever et al., 2010). These areas or topics for research are prioritised accordingly through a consensus approach. In the cardiovascular field JLA PSPs have only been conducted in the areas of cardiac surgery (Lai et al., 2020) and advanced heart failure (Taylor et al., 2020) which may be relevant to ICC patients, particularly those with severe cardiomyopathies. Neither of the top priorities for these PSPs mention anything specific for ICCs but a common theme in both includes ways in which self-management, coping and quality of life may be improved in these cardiac patients (priority 1 and 18 in the cardiac surgery PSP, and priority 1 and 3 in the advanced heart failure PSP) which is consistent with the areas covered in this research. As ICCs as a subspecialty in cardiology or genetics grows, it is likely that a JLA PSP or similar research prioritisation exercise will be conducted and thus, could

be an important consideration in the future as a starting point when choosing a research topic in the field.

9.4 Study strengths and limitations

It is important to recognise limitations of a study to clarify the extent to which it has contributed to the development of new knowledge (Ioannidis, 2007). This section considers some of the strengths and potential limitations of this research, to further support its credibility and transferability. The strengths and limitations of the systematic review and the qualitative study were discussed in Chapter 3 and Chapters 5-7, respectively. Findings from the studies should be interpreted considering these. Reflecting on the PhD project's overall strengths and limitations, this section will be discussed in the context of the application of MRC Framework in the development of the intervention and the methods applied at each phase of the research.

9.4.1 The application of the Medical Research Council framework in the study design

The strengths of the MRC framework to the research design included the ability to undergo an iterative process by which to develop an evidence-based complex intervention. The scope of the PISICC project is in the Development stage of the MRC framework, which is considered the most important phase in intervention design. By following the steps in the Development stage, this allowed the PhD research to build upon the evidence of the individual studies.

The first step of the Development stage is identifying the evidence base and as there were no psychoeducational interventions specific to family members undergoing cardiac screening/PGT at the start of the PhD project, this prompted Phase 1 of the project which consisted of the systematic review which provided a comprehensive account of the experiences, impact, and preferences of family members at risk for an

ICC undergoing cardiac screening and/or PGT. By synthesising quantitative and qualitative studies in this mixed methods review, the targets, components and outcome measures for the intervention were identified however, the form and manner for delivering the intervention was not elicited. Building on from this evidence, a qualitative study was conducted in Phase 2 of this project which strengthened the findings of the systematic review and gave greater insights on the form and delivery of the intervention.

Identifying and developing theory is a distinct second step in the Development stage of the MRC framework, however, in this project, this was a continuous process alongside each phase to ensure consistency and applicability of SDT as a theoretical basis for developing the intervention. Themes central to SDT consistently emerged in the analysis of the systematic review and the qualitative study. This therefore enabled the development of a logic model from which an intervention prototype was proposed.

Modelling process and outcomes is the final step in the Development stage of the MRC framework, and this corresponded to Phase 3 of this PhD project where a modified consensus approach was conducted with the PPI group to ensure an appropriate and relevant intervention prototype, delivery method and outcome measures. This robust iterative process coupled with the recommendation of the MRC framework to clarify the decision-making process in each phase, helped in taking a reflective and critical approach to intervention development.

The limitations of the MRC framework within this PhD project lies in its application. There are many studies citing the MRC framework as guidance for the development of complex interventions but examples in which the step-by-step application of the framework was described are rare (Hawe, 2015). A systematic review of complex nursing intervention development has identified that despite the use of the MRC

framework, studies lacked attention to psychological and social dimensions during the modelling process, there were difficulties in developing a standardised intervention and there was a lack of reporting of outcomes related to environmental factors and the context within which the interventions were delivered (Pinto et al., 2018). Whilst adherence to available guidelines (Craig et al., 2008, University of York NHS Centre for Reviews Dissemination, 2009) and a rigorous focus on the Development stage was undertaken for the PhD project, the lack of clarity over the methods to be applied to each MRC framework phase hindered fully informed decisions on the methods to use during the development of the intervention. Indeed, an update of the MRC framework is soon to be published and prior to further steps to progress to the next stage of intervention development, any new guidelines must be considered.

9.4.2 Critique of study methods

In Phase 1 of this research, the systematic review provided a comprehensive account of the experiences of at-risk family members who are undergoing cardiac screening/PGT for an ICC by synthesising quantitative and qualitative studies. A systematic review focused on the patient experience was deemed appropriate in generating the evidence base for the intervention as no specific interventions were in existence for this group. To enable the inclusion of non-experimental quantitative studies and qualitative studies, a modification of the standard guidelines in conducting the systematic review (University of York NHS Centre for Reviews Dissemination, 2009) was necessary. Measures were taken to ensure robustness in the methodology including quality appraisal and checking of themes by a second researcher and the availability of a third researcher to adjudicate any disagreements. Whilst this approach to a systematic review expands the possibilities in generating robust evidence, particularly around the phenomena of patient experience, consistency in the conduct and standard of mixed methods reviews is still evolving.

The findings of a systematic review are only as reliable as the studies included in the review and the included studies were not without their methodological issues. Although 2 of the included studies in the systematic review were deemed poor quality, no studies were excluded as very few focused solely on the experiences of at-risk family members and there was hardly any representation of those who did not take up screening and/or PGT. These issues should be considered in interpreting the findings and applying them to future studies.

In Phase 2 of the PhD research the qualitative study was justified as the systematic review did not elicit the form and manner by which the intervention should be delivered. The qualitative study was able to confirm the findings in the systematic review and gave unique insights on the perceptions, preferences, and recommendations for a psychoeducational intervention. The sampling frame ensured a breadth of patient and family experiences and ICC status. However, it is also important to consider that the psychoeducational intervention is likely to be delivered by a suitably qualified healthcare professional such as a CGN and set in a busy healthcare service with their own pressures. The researcher's own background as an experienced CGN and knowledge of the local and regional practice settings contributed to this aspect. Furthermore, healthcare professionals were part of the KHP-PPICC group that gave input into the PhD research from the beginning and their views were formally captured during the Phase 3 Modelling process for the intervention prototype. However, there would have been an advantage to capture the views of the healthcare professionals alongside the patient participants as part of the qualitative study to enhance the evidence base for the intervention. Considering the advent of methodological approaches using more integrative co-design principles, bringing patients and professionals together through a more collaborative and interactive approach, can further build on the findings in this study (McAllister et al., 2021).

Finally in Phase 3 of the PhD project, the modified consensus exercise with the PPI group was used to gather the opinions of end-users and implementers as to which components, features, outcome measures and the method of delivery of the intervention were appropriate and applicable to the population and practicable within the current healthcare setting. The consensus exercise brought about refinements to the intervention to generate an intervention prototype and recommendations for outcome measures. This fulfilled the Modelling process and outcomes step and completed the scope of the PhD project within the MRC framework. More importantly, the PPI activities conducted all throughout the research have contributed greatly to a patient-centred approach to developing health care interventions.

9.4.3 Critique of theory

The application of self-determination theory as the basis of the intervention was an iterative process starting with the themes emerging from the systematic review in Chapter 3 that corresponded to the SDT's central tenets of basic psychological needs satisfaction (autonomy, competence, and relatedness). A critical exploration of theories applicable to genetic and cardiovascular healthcare in Chapter 4 helped justify the novel application of SDT to the ICC field as it is at the forefront of providing empirical evidence based on clinical trials of SDT-based interventions as well as providing a unifying theoretical framework incorporating an individual's social context, a key aspect in inherited diseases. SDT was further tested for applicability in genetic healthcare through the qualitative study. SDT themes were not imposed during the initial coding of data and applied only to the latter stages of the Framework method of analysis which allowed SDT concepts to emerge from the data.

Self-determination theory is a theory of motivation, in which the satisfaction of the basic psychological needs brings about the highest form of engagement and intrinsic

motivation (Ryan and Deci, 2017). Developing this PhD project with SDT incorporated in all phases was valuable to understand how people adjust and cope with their journey from at-risk relative to undergoing cardiac screening and/or PGT then adjusting to the results and dealing with the impact on their families. Its use was therefore appropriate for underpinning an intervention which requires recognition of the needs for knowledge, skills, and support to make autonomous health decisions within a social context, to decrease anxiety and promote coping.

In the Modelling phase, SDT was beneficial in developing the logic model for the intervention with its constituent inputs and components, mechanism of action and outcome measures. It could be argued that a combination of theories need to be used in developing complex interventions (Pinto et al., 2018), however, most SDT based interventions are complex interventions which have been subjected to RCTs. Coupled with the strong theory-driven research design in the PhD, what remains is to test the SDT-based intervention model developed through this research through feasibility/piloting to gain more insight into the path analysis for causality. The PhD research has expanded the evidence of SDT and opened the possibility of its use in genetic healthcare.

9.5 Implications for clinical practice

This study has identified a range of areas from which the care of family members at-risk for an ICC can be improved. The implications for clinical practice for each constituent study in this research were detailed in Chapter 3 and Chapter 6. In assessing the implications of the PhD project, it is worthwhile emphasising:

There should be an emphasis that engagement in an ICC service does not automatically result in cardiac screening/PGT to enable patients to make informed choices even if they decide not to go ahead with tests.

A dominating theme in this research is that sudden cardiac death and being a parent is an important factor in the motivations and healthcare decision-making of ICC families and exploration of these aspects and support with grief is crucial prior to cardiac screening/PGT.

Patients' families are their foremost concern and the needs surrounding these will need to be addressed prior to or concurrent with the patients' individual care.

Patients prefer to be seen in a specialised ICC clinic; however, the tertiary centres will need to develop networks and pathways within their regions to ensure patients received joined up care when they need to be seen more locally.

Patients value the relationships they have with their clinicians and plays a role in their adjustment and coping but due to complexity in their screening/diagnostic journeys, simple communication issues such as not having all their information available or inconsistent advice can cause this relationship to break down.

Patients prefer family-centred care as support and learning how to cope with ICCs is also fostered through this social group. This is not counter to a more tailored approach to individual concerns particularly around lifestyle advice and symptom management.

9.6 Implications for future research

The findings of this PhD project have highlighted other areas that require further exploration in relation to enhancing the care of patients and families affected by ICCs. The implications for further research for each constituent study in this research was detailed in Chapter 3 and Chapter 7. Based on the PhD project as a whole, the implications for research are:

As a group, at-risk family members for ICCs are a distinct and growing population. There is a subset of patients who are diagnosed with an ICC and remain asymptomatic or are asymptomatic carriers; and many who do not engage with healthcare services. This group and sub-groups require focused prospective studies to ascertain their specific needs and to support the development of appropriate interventions to address these. This also applies to research around family communication for cascade screening wherein the focus is mainly on the probands and have not considered the views of the receivers of the communication who in turn, determine the outcome of these interventions in the form of uptake of screening/PGT.

Sudden cardiac death and bereavement has been highlighted to have a great impact on families in this research. However, the specific support and interventions required by those affected is largely unexplored.

As cardiac screening and/or PGT become more widely available, it is important to include diverse populations in future studies to capture a greater breadth of patient experiences. Currently, most research is carried out in Australia. North America and Europe involving mainly Caucasian participants or ethnicity is poorly described. Indeed, in this study the majority of participants were mainly White-British and were in professional job roles.

In terms of taking forward this PhD project to progress to the next phases of the MRC Framework, the intervention prototype would need to undergo feasibility testing prior to being subjected to a full-scale RCT. This is to ensure that the components, features, delivery methods and outcome measures for the intervention are appropriate and acceptable to patients and their families; and, to support the design of a future definitive trial. It is likely that modifications will be required following feasibility testing

as such is the emphasis of the MRC Framework on feedback loops across the different phases of intervention development and evaluation to bring about a robust complex intervention.

9.7 Conclusion

This PhD study provides an original contribution to the body of knowledge on the development of interventions to support patients and families affected by ICCs. This study adds to the existing evidence base on the experiences of family members at-risk for an ICC as they undergo screening and/or PGT and provided greater insight on the psychological impact of sudden cardiac death, how families coach each other in ICC care and other support systems utilised. Studies in this PhD have justified and satisfied the primary need to develop a theory-driven psychoeducational intervention to decrease anxiety and promote coping and adjustment in those with a new diagnosis of an ICC and/or genetic carrier status following screening/PGT.

The application of SDT in this research supported the understanding of the phenomena underlying patient coping, adjustment, and growth in the context of an ICC and facilitated the identification of targets, components, features, and modes of delivery for the health education and psychological aspects of the intervention. This research is a novel contribution to the scope of SDT with the expansion into genetic healthcare.

The involvement of the PPI group all throughout the PhD research from inception has contributed greatly to the applicability of all aspects of the research to end-users and through the formal consensus exercise have developed the prototype of the first of its kind psychoeducational intervention model in the field of ICCs that can be subjected to initial feasibility testing.

9.8 Dissemination and awards

2021

- Euroheartcare (Online), June 18-19, 2021

Bueser, T., Patch, C., Rowland, E., Coles, L. and Metcalfe, A., 2021. Patient & public involvement for inherited cardiac conditions. European Journal of Cardiovascular Nursing, 20(Supplement_1), pp.zvab060-086.

2019

- Early Career Researcher Award, Cardiovascular South London Clinical Research Network, November 2019

- Poster prize winner: World Congress on Genomic Counselling (Cambridge UK), October 2-4, 2019

Bueser, T., Patch, C., Rowland, E., Carr-White, G. Metcalfe, A., A psychoeducational intervention supporting patients with a new diagnosis and/or genetic carrier status for an inherited cardiac condition (PISICC)

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Appendix 1 Terms of reference for the King's Health Partners' Patient & Public Involvement for Inherited Cardiac Conditions (KHP-PPIICC) group

Name of group: King's Health Partners 'Patient & Public Involvement for Inherited Cardiac Conditions (PPIICC)

Title: Terms of reference (March 2017)

Purpose / role of the group:

The role of the Group is to advise on the development and coordination of Patient and Public Involvement (PPI) in the doctoral research activities of Tootie (Teofila) Bueser and service provision of the King's Health Partners 'Inherited Cardiac Conditions (ICC) clinic. The Group will act as a 'critical friend 'and provide guidance to ensure that research and clinical services are delivered to a high standard, remain patient and family-centred and ethically sound.

Membership:

The group will be comprised of individuals who are patients or family members/carers of those affected by an ICC who wish to be involved in clinical research but not necessarily as research participants. They will be an advisor for the researcher and clinical staff, help review literature such as patient information sheets and proposals; and assess patient pathways.

Members will be expected to:

- make a reasonable contribution of time to attend meetings
- make an effort to read through information sent in advance of meetings
- offer constructive feedback and take an active role within group discussions

- make honest mileage and expenses claims, using original receipts where possible.

- inform the researcher prior to meetings if they are unable to attend.

The group will:

- respect anonymity and confidentiality of discussion

- consider all representative views

- determine areas of particular priority for discussion

- be informed of the research topics prior to the meeting

- be informed of the attendance of specialists / researchers prior to the meeting

Any reasonable travel costs to the lay members will be reimbursed by the researcher. Group members will also be compensated for time spent attending meetings.

In recognition of data protection, members who use their personal emails will be blind carbon copied to protect their personal details.

Group membership is provisionally held until May 2019.

Working methods / ways of working:

- The Group will meet a minimum of four times a year (up to a maximum of 6 times per year)

- Members of the Group will receive papers one week before each meeting.

- Minutes of the meeting will be kept by a minutes secretary and agreed by all members of the Group who attended the meeting.

- Members may be contacted between meetings for advice should the need arise.

- From time-to-time sub-groups may be formed to work on specific issues as appropriate.
- From time-to-time individuals may be co-opted to provide specific advice and expertise as required.

Sharing information:

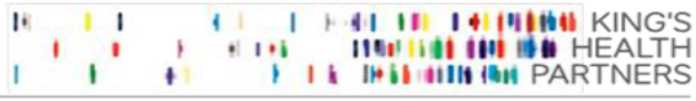
Information sharing will primarily be though email contact, unless specified differently. Lengthy meeting papers should be sent with no less than 1 week notice and are available as electronic or paper copies. Short documents (<2 pages) can be made available on the day of the meeting although efforts to send them prior to the meeting should be made.

Review:

The terms of reference (TOR) will be initially reviewed with the Group members and thereafter the group will review the TOR annually.

Appendix 2 Study documents

A. Recruitment poster



An Academic Health Sciences Centre for London — Pioneering better health for all

****RESEARCH PROJECT RECRUITMENT****

Helping families with Inherited Heart Conditions

Study Title: Developing a psychoeducational intervention to improve the uptake of cardiac screening and/or predictive genetic testing for inherited cardiac conditions

Can you help?
I am looking for patients who would like to take part in a group discussion or one-on-one interview in order to know more about the experiences of people at risk for inherited heart conditions for a PhD research project.

Who can take part?
I would like to talk to patients referred for cardiac screening or genetic testing for a family history of-

an inherited type of cardiomyopathy:

- hypertrophic
- dilated
- arrhythmogenic

OR

an inherited heart rhythm disorder:

- long qt syndrome
- brugada syndrome
- catecholaminergic polymorphic ventricular tachycardia

How can I take part?
If you are interested in taking part in the study, please approach your **cardiac genetics specialist nurse** or contact:

Teofila (Tootie) Bueser
Telephone number: 07738705554
Email: tootie.bueser@kcl.ac.uk

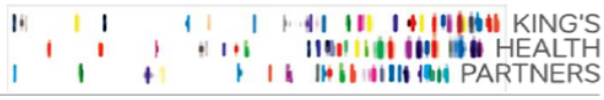
Thank you!

Version 1.1 December 5 2016

IRAS Project ID: 209714

KING'S College LONDON — Guy's and St Thomas' NHS — King's College Hospital NHS — South London and Maudsley NHS

B. Participant information sheet



An Academic Health Sciences Centre for London — Pioneering better health for all

Participant Information Sheet
Version 1.1 December 5, 2016

Study Title: Developing a psychoeducational intervention to improve the uptake of cardiac screening and/or predictive genetic testing for inherited cardiac conditions

We would like to invite you to take part in our research study. Before you decide, we would like you to fully understand why the research is being done and what your involvement will be. A member of our team will go through this information sheet with you and can answer any questions you may have.

What is the study about? The study will look into the experiences of patients undergoing screening for an inherited heart condition.

Why have I been approached to participate in this study? You have been approached to participate in this study because you have been referred for family screening in the Inherited Cardiac Conditions clinic at King's College Hospital or Guy's & St Thomas' Hospital. Your genetic counsellor, specialist nurse or heart doctor has informed us that you may be eligible for this study.

Do I have to take part? No, you may choose whether to participate in the study or not. Your decision will not affect your future treatment or care.

What do I do if I want to take part? If you are interested in taking part in the study, please inform your genetic counsellor, specialist nurse or heart doctor who will pass on your contact details to the researcher. Alternatively, you may contact the researcher directly to discuss your participation in the study.

What will I have to do? The researcher will schedule one group discussion or if you prefer, a face-to-face interview at King's College London or in your home lasting approximately 60 minutes. It will be conducted by a trained researcher and recorded using a digital voice recorder.

What will happen during the group discussion or interview? You will be asked to share your experience and views of finding out about the risk for an inherited heart condition and what you felt was useful/not useful in helping you make decisions about your care.

What will happen to my personal information and the recorded information? Any personal information that you share or obtained by us from your medical records (such as your contact details, family history and cardiac or genetic test results) and the recordings from the group discussion or interviews will be strictly confidential. Steps to protect the identity of each person taking part include assigning a code that will replace their name and other personal details so that their data will be anonymous to everyone except the researcher. Furthermore, after the voice recordings have been made into

typed documents, the original recording will be destroyed and all personal information will be removed. We will also ensure that no individuals will be identifiable in all published papers and quotations taken from the interviews will be anonymised.

Any documents and data from the research will be stored securely in password protected files on computers at King's College London.

What are the benefits? There will be no direct benefits to you or your family. However, individuals taking part in similar studies have found it a positive experience to talk about being tested for a genetic condition and how they coped with the results.

What are the risks? All of the questions have been carefully designed so that they do not cause stress to you. However, there may be sensitive issues or rare circumstances that the researcher is not aware of, or something you find distressing is discussed. Should you become upset, we can discuss whether you want to stop or take a break from the interview or if you need further support from your clinical team.

Will I receive any compensation? You will be reimbursed in the form of vouchers for reasonable transport costs for attending the focus group or if the interview was not conducted in your own home. You can also claim for childcare vouchers if you needed childcare services to enable you to participate in this study. Claim forms will be provided when the focus group or interview is scheduled.

What happens if I change my mind during the study? If you decide that you no longer want to participate in the study, please let the researcher know. You will not be obliged to give a reason for your withdrawal from the study and it will not affect your care or treatment in the future.

What happens at the end of the study? The findings of this study will be used to develop an intervention to support patients with decision-making as they go through heart tests and will be published in academic journals. We will provide an overview of the findings at the end of the study in a feedback session and through a written summary. Please inform the researcher if you prefer not to attend the session or receive the summary.

What happens if I have any concerns or a complaint? If you have a concern or a complaint about the study please contact the Chief Investigator who will be happy to answer your questions:

Chief investigator: Professor Alison Metcalfe
Address: Florence Nightingale Faculty of Nursing & Midwifery, King's College London, 57 Waterloo Road, London SE1 8WA
Telephone: 020 7848 3828
E-mail: alison.metcalfe@kcl.ac.uk

If you remain unhappy, you may be able to make a formal complaint through the NHS Complaints Procedure. Details can be obtained through the KCH PALS (Patient Advice and Liaison service) department. Their contact details are:

Address: PALS, King's College Hospital, Hambleden Wing Central, Denmark Hill, London, SE5 9RS
Telephone: 02032993601

Who organised and funded the study? The study is being sponsored by King's College London and co-sponsored King's College Hospital NHS Foundation Trust. The study is being conducted as part of a PhD degree and is funded by a fellowship grant from the National Institute for Health Research.

Who has reviewed the study? This study was given favourable ethical approval by the London-Fulham Research Ethics Committee (17/LO/0059).

What if I have more questions or do not understand something? You may have several questions about the study that you might want to ask before you agree to take part in the interview. Please contact the researcher who will be happy to answer your questions:

Researcher: Teofila (Tootie) Bueser
Address: Florence Nightingale Faculty of Nursing & Midwifery, King's College London, 57 Waterloo Road, London SE1 8WA
Telephone: 07738705554
E-mail: tootie.bueser@kcl.ac.uk

C. Participant consent form

KING'S HEALTH PARTNERS
An Academic Health Sciences Centre for London — Pioneering better health for all

PARTICIPANT CONSENT FORM

Project Title: Developing a psychoeducational intervention to improve the uptake of cardiac screening and/or predictive genetic testing for inherited cardiac conditions

Version 1.1 05/12/2016

Please initial boxes

1. I confirm that I have read and understood the information leaflet dated __/__/__ (version_) for the study: Developing a psychoeducational intervention to improve the uptake of cardiac screening and/or predictive genetic testing for inherited heart conditions. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily. ☐
2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving any reason and without my medical care or legal rights being affected. ☐
3. I understand that relevant sections of my medical notes and data collected during the study, may be looked at by individuals from the project or from the NHS Trust, where it is relevant to my taking part in this research. I give permission for these individuals to have access to my record. ☐
4. I agree to participate in a group discussion with up to 6 participants with similar experiences or have a face-to-face interview lasting for about 1 hour. ☐
5. I understand that the group discussions or interview will be audio-recorded. ☐
6. I understand that should I feel uncomfortable or upset during the discussion or interview, I can request for the session to be discontinued and I will be offered additional help and advice from the clinical team looking after me for my condition. ☐
7. I understand that the recordings will be transcribed word for word and analysed. The results of the study will be published and direct quotes may be printed but my identity will be kept confidential. ☐
8. In the event that in the course of the discussions, it is apparent that I need additional input or modification in my clinical management, I agree that my clinical care team or GP can be notified ☐
9. I understand I will be reimbursed in the form of vouchers for reasonable transport costs and childcare expenses to enable me to participate in the discussions. ☐
10. I understand that if I have any questions or concerns, the main study contact is: ☐

    Teofila (Tootie) Bueser
    Telephone: +44 773 870 5554
    Email: tootie.bueser@kcl.ac.uk

Optional Feedback of results

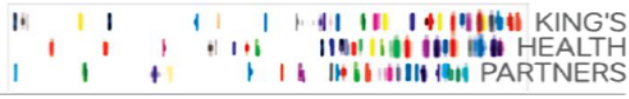
11. I give consent for the research team to feedback the analysis of the study to me. ☐
12. I would like to participate in a further group discussion lasting for one hour for the feedback of the results. ☐

Name of Participant (BLOCK CAPITALS)	Date of Birth	Date	Signature

Name of Researcher (BLOCK CAPITALS)	Date	Signature

Original to be kept by researcher, one copy to be kept in the medical notes and a copy to be given to participant

D. Topic guide



An Academic Health Sciences Centre for London — Pioneering better health for all

Topic guide for focus groups/interviews
Version 1.1 December 5, 2016, IRAS Project ID: 209714

Title of Project: Developing a psychoeducational intervention to improve the uptake of cardiac screening and/or predictive genetic testing for inherited cardiac conditions

Introduction: Welcome to the session. As part of my PhD degree at King's College London, I am conducting focus group discussions or interviews with patients at risk for inherited heart conditions. The information we gather today will be used to help write a research report on the experiences of patients and to help develop a psychoeducational tool to help support this patient group in the future. Please be reassured that no individual will be named in our report and nothing will be linked back to any participant. Our discussion should take around 1 hour and will be audio-recorded using an encrypted digital recorder.

Are you happy to participate in the focus group or interview today? You are free to withdraw from the discussion at any time and you do not have to give a reason. Reimbursement for your transport and childcare costs will be organised at the end of the session. Please refrain from using electronic devices.

I would like to emphasise that there are no right or wrong answers and all your opinions are important for this study in order to understand your experiences and incorporate this in the psychoeducational intervention we are developing. (**FOR FOCUS GROUPS:** As there are a couple of you in this group and you may have differing opinions, please listen and respond respectfully as others share their views and feel free to share your point of view even if it differs from what others say.) The expectation is that anything said within the group or the interview remains confidential but if you are concerned about giving personal information that you do not want others to know, you do not have to share it. Are there any questions before we start?

Topics

1. **Introductions from the participant/s**

   -name
   -share an interest or a hobby

2. **Genetic risk for an inherited heart condition**

   -concept of genetic risk
   -source of risk information
   -thoughts and feelings

3. **Exploring support for Self-determination in decision-making**

   3.1 Relatedness

   3.2 Autonomy

   3.3 Competence

   -facilitators and barriers
   -role of and relationships with family, support networks and clinical team
   -information resources
   -mapping prioritisation, decision-making, motivation

4. **Factors that may improve experience of screening / genetic testing**

   -resources
   -structure / pathways
   -clinical staff attributes
   -support system attributes

5. **Explore the most valuable resource to support experience of screening / genetic testing**

6. **Questions from the group, if any.**

Conclusion: Thank you very much for your time and for sharing your thoughts and experiences today. You will receive a written summary of the findings once the analysis is completed. In addition, an invitation for a feedback session will be sent to who those have indicated that they would like to come for a further session.

Appendix 3 Example of a sample search strategy used in Medline (Ovid)

# ▲	Searches	Results
1	(Predictive adj3 (genetic or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	2024
2	(Pre?symptomatic adj (genetic or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	333
3	(cascade adj3 (screening or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	363
4	(family adj3 (screening or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	2472
5	(genetic adj3 counsel*).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	20761
6	inherited.mp.	60964
7	genetic.mp.	1322912
8	cardiac.mp.	615864
9	heart.mp.	1047729
10	cardiovasc*.mp.	427809
11	arrhythmias.mp. or Arrhythmias, Cardiac/	81968
12	Cardiomyopathy, Dilated/ or Cardiomyopathies/ or Arrhythmias, Cardiac/ or cardiomyo*.mp. or Cardiomyopathy, Hypertrophic/	153708
13	(arrhythmogenic adj3 (right or cardiomyo*)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	2713
14	(Predictive adj3 (genetic or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms]	2024

15 (Pre?symptomatic adj (genetic or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 333

16 (cascade adj3 (screening or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 363

17 (family adj3 (screening or testing)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 2472

18 (genetic adj3 counsel*).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 20761

19 inherited.mp. 60964

20 genetic.mp. 1322912

21 cardiac.mp. 615864

22 heart.mp. 1047729

23 cardiovasc*.mp. 427809

24 arrhythmias.mp. or Arrhythmias, Cardiac/ 81968

25 Cardiomyopathy, Dilated/ or Cardiomyopathies/ or Arrhythmias, Cardiac/ or cardiomyo*.mp. or Cardiomyopathy, Hypertrophic/ 153708

26 (arrhythmogenic adj3 (right or cardiomyo*)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 2713

27 (long qt adj3 syndrome).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 7989

28 (catecholaminergic adj3 (polymorphic or ventricular or tachycardia)).mp. [mp=title, abstract, original title, name of substance word, subject heading word, keyword heading word, protocol supplementary concept word, rare disease supplementary concept word, unique identifier, synonyms] 628

29 Brugada Syndrome/ or brugada.mp. 4086

30 14 or 15 or 16 or 17 or 18

31 19 or 20 1355332

32 21 or 22 or 23 or 24 or 25 or 26 or 27 or 28 or 29 1575763

33 31 and 32 57338

34 30 and 33 1510

Appendix 4 Summary of quality assessment for the mixed-methods Systematic review

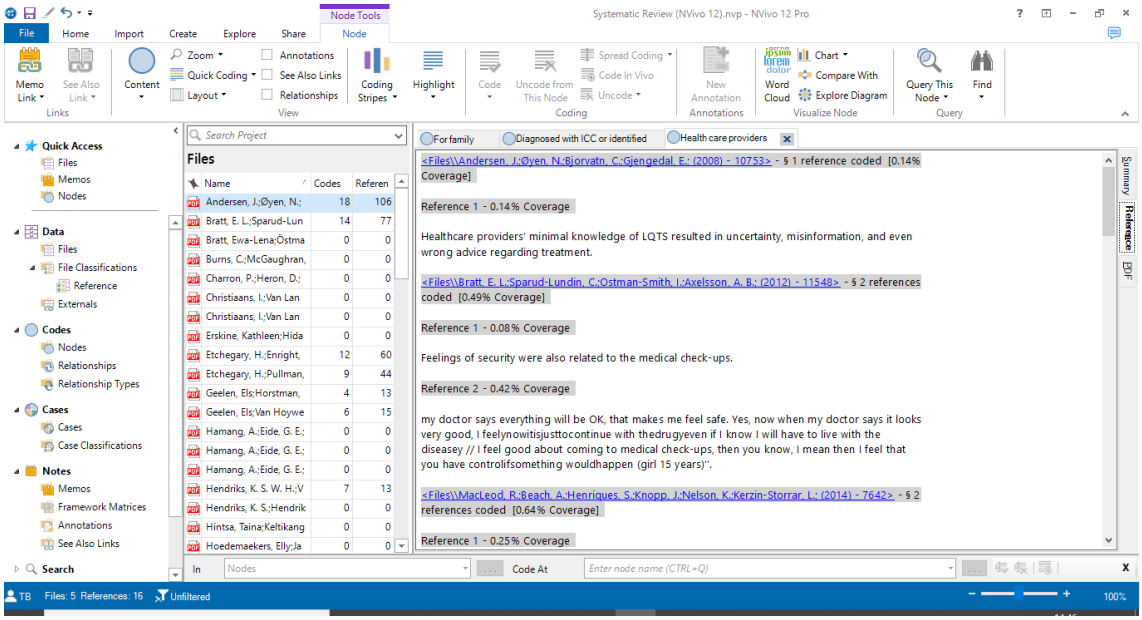
Study design	Quantitative															
Study number	**1**	**2**	**3**	**4**	**5**	**6**	**7**	**8**	**9**	**10**	**11**	**12**	**13**	**14**	**15**	**16**
References	**Bratt, Osman-Smith et al. (2012)**	**Charron et al. (2002)**	**Christiaans et al. (2009)**	**Christiaans et al. (2008)**	**Hamang et al. (2010)**	**Hamang et al. (2011)**	**Hamang et al. (2012)**	**Hendriks et al. (2008)**	**Hintsa et al. (2009)**	**Hoede maekers et al (2007)**	**Ingles et al. (2008)**	**Ingles et al. (2012)**	**Jensen et al. (2013)**	**Khouzam (2015)**	**McGorrian et al. (2013)**	**Smets et al. (2008)**
Question/objective sufficiently described?	2	1	2	2	2	2	2	1	2	1	2	2	1	2	2	2
Study design evident and appropriate?	1	0	2	2	1	1	2	1	1	2	1	2	1	2	2	2
Context for the study clear?	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2
Sampling strategy described, relevant and justified?	1	0	1	1	2	2	2	2	1	2	1	2	2	1	1	2
Participant group/s adequately described?	2	2	2	2	2	2	2	2	1	1	2	2	2	2	2	2
Conclusions supported by the results?	2	1	2	2	2	2	2	2	2	2	2	2	1	2	2	2

Study design	Quantitative															
Study number	**1**	**2**	**3**	**4**	**5**	**6**	**7**	**8**	**9**	**10**	**11**	**12**	**13**	**14**	**15**	**16**
References	**Bratt, Osman-Smith et al. (2012)**	**Charron et al. (2002)**	**Christiaans et al. (2009)**	**Christiaans et al. (2008)**	**Hamang et al. (2010)**	**Hamang et al. (2011)**	**Hamang et al. (2012)**	**Hendriks et al. (2008)**	**Hintsa et al. (2009)**	**Hoede maekers et al (2007)**	**Ingles et al. (2008)**	**Ingles et al. (2012)**	**Jensen et al. (2013)**	**Khouzam (2015)**	**McGorrian et al. (2013)**	**Smets et al. (2008)**
Data collection methods clearly described and systematic?	2	0	2	2	1	1	2	1	1	2	1	1	1	2	1	2
Data analysis clearly described and systematic?	2	1	2	1	2	2	2	2	2	1	2	2	0	1	2	2
Sampling size adequate? (Representativeness of both participant groups? Clustering for clinician?)	0	0	0	0	1	1	1	1	1	1	0	0	0	0	0	0
Measurements likely to be valid and reliable	1	0	1	1	2	2	2	2	1	1	2	2	2	1	2	1
Total	15	7	16	15	17	17	19	16	14	15	15	17	12	15	16	17
Score	75%	35%	80%	75%	85%	85%	95%	80%	70%	75%	75%	85%	60%	75%	80%	85%

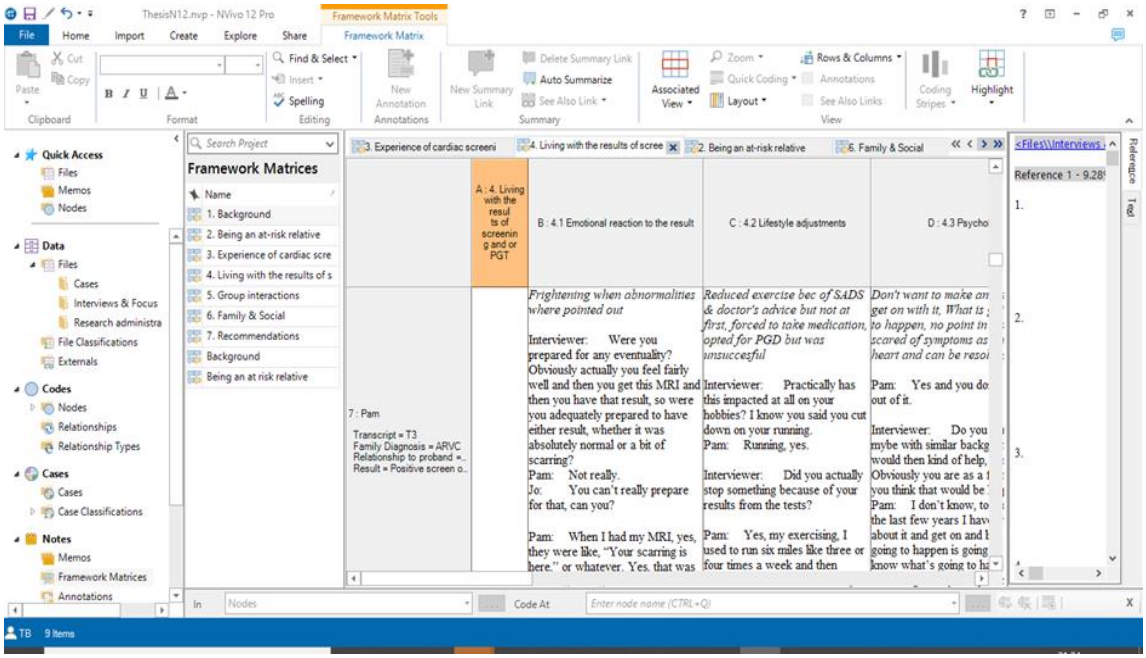
Study design	Qualitative														Mixed methods
Study number	**17**	**18**	**19**	**20**	**21**	**22**	**23**	**24**	**25**	**26**	**27**	**28**	**29**	**30**	**31**
References	**Andersen et al. (2008)**	**Bratt et al. (2012)**	**Etchegary et al. (2015)**	**Etchegary et al. (2016)**	**Geelen et al. (2011)**	**Geelen et al (2012)**	**MacLeod et al. (2014)**	**Manuel & Brunger (2014)**	**Manuel & Brunger (2015)**	**Meulenkamp et al. (2008)**	**Ormondroyd et al. (2014)**	**Smart (2010)**	**van der Werf et al. (2014)**	**Whyte et al. (2016)**	**Hendriks et al. (2005)**
Question/objective sufficiently described?	2	2	1	1	2	2	2	1	1	2	2	1	2	2	2
Study design evident and appropriate?	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2
Context for the study clear?	2	2	2	2	2	2	2	2	2	2	2	2	2	2	2
Sampling strategy described, relevant and justified?	2	2	2	2	2	2	2	2	2	2	2	2	2	1	0
Participant group/s adequately described?	2	2	2	2	1	1	2	2	2	2	2	1	2	1	1
Conclusions supported by the results?	2	2	2	2	2	2	2	2	2	2	2	2	2	2	1

Study design	Qualitative														Mixed methods
Study number	**17**	**18**	**19**	**20**	**21**	**22**	**23**	**24**	**25**	**26**	**27**	**28**	**29**	**30**	**31**
References	**Andersen et al. (2008)**	**Bratt et al. (2012)**	**Etchegary et al. (2015)**	**Etchegary et al. (2016)**	**Geelen et al. (2011)**	**Geelen et al (2012)**	**MacLeod et al. (2014)**	**Manuel & Brunger (2014)**	**Manuel & Brunger (2015)**	**Meulenkamp et al. (2008)**	**Ormondroyd et al. (2014)**	**Smart (2010)**	**van der Werf et al. (2014)**	**Whyte et al. (2016)**	**Hendriks et al. (2005)**
Data collection methods clearly described and systematic?	2	2	2	2	1	1	2	1	1	2	2	2	2	2	0
Data analysis clearly described and systematic?	2	2	1	2	1	1	2	2	2	2	2	2	2	2	0
Connection to a theoretical framework/wider body of knowledge?	1	1	1	1	1	1	1	2	2	2	1	1	1	1	0
Use of verification procedure(s) to establish credibility? (e.g. co-coding, reflexivity, data triangulation)	1	2	1	2	2	2	2	2	2	2	2	1	2	0	0
Total	18	19	16	18	16	16	19	18	18	20	19	16	19	15	10
Score	90%	95%	80%	90%	80%	80%	95%	90%	90%	100%	95%	80%	95%	75%	42%

Appendix 5 Systematic review analysis sample



Appendix 6 Qualitative study framework analysis matrix

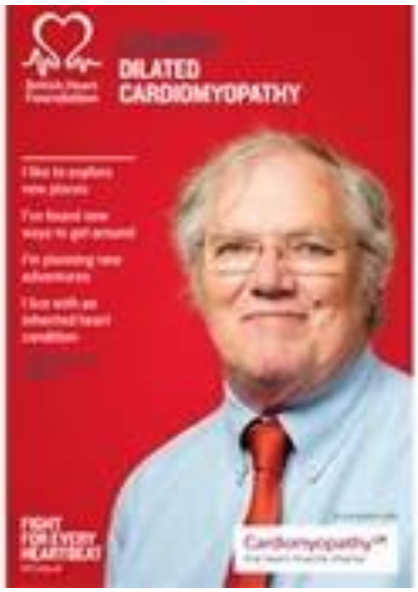


Appendix 7 Psychoeducational intervention materials

A. Information leaflets

Dilated cardiomyopathy

(https://www.bhf.org.uk/informationsupport/publications/heart-conditions/life-with-dilated-cardiomyopathy)

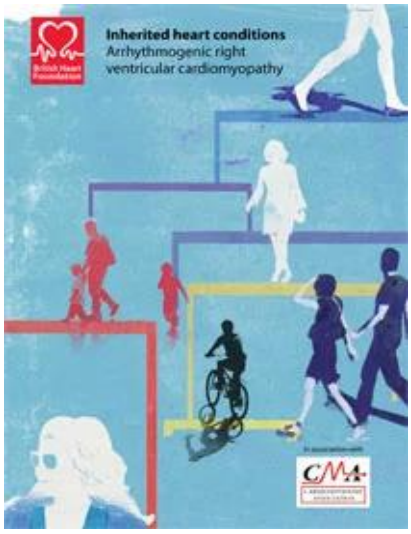


Hypertrophic cardiomyopathy

https://www.bhf.org.uk/informationsupport/publications/heart-conditions/m111c-inherited-heart-conditions---hypertrophic-cardiomyopathy)

Arrhythmogenic right ventricular cardiomyopathy

(https://www.bhf.org.uk/informationsupport/publications/heart-conditions/m111e-inherited-heart-conditions---arrhythmogenic-right-ventricular-cardiomyopathy)



Inherited abnormal heart rhythms

(https://www.bhf.org.uk/informationsupport/publications/heart-conditions/m111b-inherited-heart-conditions---inherited-heart-rhythm-disturbances)

B. Personalised lifestyle form based on the European Society for Cardiology Hypertrophic Cardiomyopathy Guidelines

Lifestyle aspect	Changes that already happened since my diagnosis or genetic carrier status	Changes that I feel need to be made as a result of my diagnosis or genetic carrier status (or questions)	I don't think any changes are needed (√)
Exercise			
Diet, alcohol, weight			
Smoking			
Sexual activity			
Medication			
Vaccination			
Driving			

Lifestyle aspect	Changes that already happened since my diagnosis or genetic carrier status	Changes that I feel need to be made as a result of my diagnosis or genetic carrier status (or questions)	I don't think any changes are needed (√)
Occupation			
Holidays & travel insurance			
Life insurance			
Pregnancy & childbirth (or starting a family)			

Appendix 8 Outcome questionnaires

A. Perceived competence for inherited heart conditions

Please respond to each of the following items in terms of how true it is for you with respect to dealing with your diagnosis or carrier status for an inherited heart condition.

1. I feel confident in my ability to manage my diagnosis or carrier status for an inherited heart condition.

1	2	3	4	5	6	7
not at all true			somewhat true			very true

2. I am capable of handling my diagnosis or carrier status for an inherited heart condition now.

1	2	3	4	5	6	7
not at all true			somewhat true			very true

3. I am able to do my own routine for my diagnosis or carrier status for an inherited heart condition now.

1	2	3	4	5	6	7
not at all true			somewhat true			very true

4. I feel able to meet the challenge of managing my diagnosis or carrier status for an inherited heart condition.

1	2	3	4	5	6	7
not at all true			somewhat true			very true

B. Perceived choice and awareness of self scale

Instructions: Please read the pairs of statements, one pair at a time, and think about which statement within the pair seems more true to you at this point in your life. Indicate the degree to which statement A feels true, relative to the degree that Statement B feels true, on the 5-point scale shown after each pair of statements. If statement A feels completely true and statement B feels completely untrue, the appropriate response would be 1. If the two statements are equally true, the appropriate response would be a 3. If only statement B feels true And so on.

1. A. I always feel like I choose the things I do.

   B. I sometimes feel that it's not really me choosing the things I do.

Only A feels true 1 2 3 4 5 **Only B feels true**

2. A. My emotions sometimes seem alien to me.

   B. My emotions always seem to belong to me.

Only A feels true 1 2 3 4 5 **Only B feels true**

3. A. I choose to do what I have to do.

   B. I do what I have to, but I don't feel like it is really my choice.

Only A feels true 1 2 3 4 5 **Only B feels true**

4. A. I feel that I am rarely myself.

   B. I feel like I am always completely myself.

Only A feels true 1 2 3 4 5 **Only B feels true**

5. A. I do what I do because it interests me.

   B. I do what I do because I have to.

Only A feels true 1 2 3 4 5 **Only B feels true**

6. A. When I accomplish something, I often feel it wasn't really me who did it.

B. When I accomplish something, I always feel it's me who did it.

Only A feels true 1 2 3 4 5 **Only B feels true**

7. A. I am free to do whatever I decide to do.

B. What I do is often not what I'd choose to do.

Only A feels true 1 2 3 4 5 **Only B feels true**

8. A. My body sometimes feels like a stranger to me.

B. My body always feels like me.

Only A feels true 1 2 3 4 5 **Only B feels true**

9. A. I feel pretty free to do whatever I choose to.

B. I often do things that I don't choose to do.

Only A feels true 1 2 3 4 5 **Only B feels true**

10. A. Sometimes I look into the mirror and see a stranger.

B. When I look into the mirror I see myself.

Only A feels true 1 2 3 4 5 **Only B feels true**

C. Health care climate questionnaire

This questionnaire contains items that are related to your encounters with your clinicians (doctors and cardiac genetic nurses) in the inherited cardiac conditions clinic. Clinicians have different styles in dealing with patients, and we would like to know more about how you have felt about your encounters with them. Your responses are confidential. Please be honest and candid.

Questions focused on your experience with your doctor:

1. I feel that my doctor has provided me choices and options.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

2. I feel understood by my doctor.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

3. I am able to be open with my doctor at our meetings.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

4. My doctor conveys confidence in my ability to make changes.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

5. I feel that my doctor accepts me.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

6. My doctor has made sure I really understand about my condition and what I need to do.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

7. My doctor encourages me to ask questions.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

8. I feel a lot of trust in my doctor.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

9. My doctor answers my questions fully and carefully.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

10. My doctor listens to how I would like to do things.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

11. My doctor handles people's emotions very well.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

12. I feel that my doctor cares about me as a person.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

13. I don't feel very good about the way my doctor talks to me.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

14. My doctor tries to understand how I see things before suggesting a new way to do things.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

15. I feel able to share my feelings with my doctor.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

Questions focused on your experience with your cardiac genetic nurse:

1. I feel that my cardiac genetic nurse has provided me choices and options.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

2. I feel understood by my cardiac genetic nurse

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

3. I am able to be open with my cardiac genetic nurse at our meetings.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

4. My cardiac genetic nurse conveys confidence in my ability to make changes.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

5. I feel that my cardiac genetic nurse accepts me.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

6. My cardiac genetic nurse has made sure I really understand about my condition and what I need to do.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

7. My cardiac genetic nurse encourages me to ask questions.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

8. I feel a lot of trust in my cardiac genetic nurse.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

9. My cardiac genetic nurse answers my questions fully and carefully.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

10. My cardiac genetic nurse listens to how I would like to do things.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

11. My cardiac genetic nurse handles people's emotions very well.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

12. I feel that my cardiac genetic nurse cares about me as a person.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

13. I don't feel very good about the way my cardiac genetic nurse talks to me.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

14. My cardiac genetic nurse tries to understand how I see things before suggesting a new way to do things.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

15. I feel able to share my feelings with my cardiac genetic nurse.

1	2	3	4	5	6	7
strongly disagree			neutral			strongly agree

D. Cardiac anxiety questionnaire

		Never	Rarely	Sometimes	Often	Always
1.	I pay attention to my heartbeat	0	1	2	3	4
2.	I avoid physical exertion	0	1	2	3	4
3.	My racing heart wakes me up at night	0	1	2	3	4
4.	Chest pain/discomfort wakes me up at night	0	1	2	3	4
5.	I take it easy as much as possible	0	1	0	2	4
6.	I check my pulse	0	1	2	3	4
7.	I avoid exercise or other physical work	0	1	2	3	4
8.	I can feel my heart in my chest	0	1	2	3	4
9.	I avoid activities that make my heartbeat faster	0	1	2	3	4
10.	If tests come out normal, I still worry about my heart	0	1	2	3	4
11.	I feel safe being around a hospital, physician, or other medical facility	0	1	2	3	4
12.	I avoid activities that make me sweat	0	1	2	3	4
13.	I worry that doctors do not believe my symptoms are real	0	1	2	3	4
When I have chest discomfort or when my heart is beating fast:						
14.	I worry that I may have a heart attack	0	1	2	3	4
15.	I have difficulty concentrating on anything else	0	1	2	3	4
16.	I get frightened	0	1	2	3	4

	Never	Rarely	Sometimes	Often	Always
17. I like to be checked out by a doctor	0	1	2	3	4
18. I tell my family or friends	0	1	2	3	4