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Scoping Review of Experience of People living with Inherited Cardiac Conditions and Congenital Heart Disease. The experience of individuals with inherited cardiac conditions and congenital heart disease and their families throughout the life course: impact, difficulties and resilience

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Royal College of Surgeons in Ireland

Scoping Review of Experience of People living with Inherited Cardiac Conditions and Congenital Heart Disease

The experience of individuals with inherited cardiac conditions and congenital heart disease and their families throughout the life course: impact, difficulties and resilience

Annette Burns, Professor Anne Hickey, Dr Frank Doyle
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Contents

Executive summary	4
Part I: Inherited Cardiac Conditions.....	6
1.1 Inheritable Cardiac Diseases Overall.....	7
1.2 Implantable Cardioverter Defibrillator Recipients.....	8
1.3 Hypertrophic Cardiomyopathy	9
1.4 Catecholaminergic polymorphic ventricular tachycardia (CPVT)	10
1.5 Brugada syndrome.....	10
1.6 Long QT Syndrome.....	11
1.7 HCM and LQTS	11
1.8 Dilated Cardiomyopathy	12
1.9 Arrhythmogenic Right Ventricular Cardiomyopathy	12
1.10 Familial Hypercholesterolemia	12
Part II: Congenital Heart Disease	14
2.1 Incidence of Congenital Heart Disease in Ireland.....	15
2.2 Increased survival	15
2.3 Transition	15
2.3.1 Transition in Ireland	16
2.3.2 Consequences of Inadequate Transition	16
2.4 Financial Burden of Illness	17
2.5 Insurability, Mortgage and Loan Applications	17
2.6 Employment.....	18
2.6.1 Employment rates.....	18
2.6.2 Early career guidance advice	19
2.6.3 Importance of employment.....	19
2.6.4 Social mobility.....	19
2.7 Exercise	19
2.7.1 Subjective experience	19
2.7.2 Peer comparisons.....	20
2.7.3 Barriers.....	20
2.7.4 Safety	20
2.7.5 Overestimating exercise capacity	20
2.7.6 Benefits	21
2.8 Family Socioeconomic Status.....	21

2.9	Romantic relationships	21
2.10	Sexual functioning.....	22
2.11	Contraception, Pregnancy and Heredity.....	22
2.12	CHD with Down Syndrome.....	23
2.13	Parents/Family	23
2.13.1	Diagnosis, Surgery, childhood and familial issues.....	23
2.13.2	Adolescence and beyond: Support helps but not overprotection.....	24
2.14	Growing-up with Congenital Heart Disease: Summarising the various life stages.....	25
2.14.1	Childhood	25
2.14.2	Adolescence	25
2.14.3	Young Adults	28
2.14.4	Adult patients.....	28
	Future research.....	29
	References	30

Introduction

This was a scoping review of the experiences of people with inherited cardiac conditions and congenital heart disease and their families. The Irish Heart Foundation is trying to investigate the issues which these individuals and their families might face throughout the life course. It is hoped that this scoping review will highlight current knowledge, inform potential future studies and also inform the supports provided by the IHF to people with inherited cardiac conditions and their families.

As stated, the aim of this report was to explore the experience of individuals with inherited cardiac conditions and their families throughout the life course. Due to the very limited literature available on inherited cardiac conditions (Part I), this report also explored the broader area of congenital heart disease (Part II), which has a lot more literature available. Part I of this report therefore identifies more specific detail in relation to common cardiac conditions which are inherited while Part II provides a summary of the (larger) literature available on congenital heart disease which includes inherited (Brugada syndrome, Long QT Syndrome and Progressive Cardiac Conduction Defect) and other cardiac conditions (i.e. not all congenital heart disease is inherited).

Some of the main themes which emerged as important in the literature were: Genetic testing and screening of family members, transition for younger patients, exercise/physical activity, stress and psychological adjustment of individual and their family, feeling normal, education concerning contraception and reproduction including genetic counselling, insurance and employment.

There is virtually no Irish research on inherited cardiac conditions and congenital heart disease and the literature on inherited cardiac conditions is in general limited. Many areas of interest in relation to the social consequences, psychological experiences and quality of life associated with inherited cardiac conditions could not be addressed by this review as they simply have not yet been addressed in the literature itself. These knowledge gaps highlight the need for further research in this area.

Research in this area might also be limited due to the sometimes invisible nature of these conditions. Not all inherited cardiac conditions are apparent from birth and in some cases the first symptom can actually be sudden death (1). In other cases a condition may go undiagnosed until a relative experiences symptoms or is lost to sudden cardiac death and it is only then that diagnosis is made following screening. Therefore for some, living with inherited cardiac conditions does not affect their lives at all in that they are unaware they have it.

The common inherited cardiac conditions included are also quite different and therefore studies tend to look at each individually rather than exploring the experiences of individuals with inherited cardiac conditions as a whole. The exception to this is individuals who have received implantable cardioverter defibrillators. These people are sometimes looked at as a group and therefore different inherited cardiac conditions can sometimes be combined in these studies.

Executive summary

- Inherited cardiac conditions represent a number of diverse conditions with varying symptoms, prognosis and impacts. These include arrhythmias, congenital heart disease, cardiomyopathy and high blood cholesterol (2).
- There is limited literature available on inherited cardiac conditions exclusively
- Some inherited cardiac conditions appear to have more of an impact on psychological adjustment and health status than others. For instance, cardiomyopathies appear to be more burdensome than familial hypercholesterolemia in this regard (3).
- Genetic testing and screening seem to be well tolerated and longitudinal studies suggest emotional distress and psychological adjustment in those at risk and who undergo testing is similar to general population levels.
- Patients who have been implanted with an implantable cardioverter defibrillator (ICD) have higher levels of post-traumatic stress disorder when compared to the general population (4, 5). Most do adjust well but researchers recommend all device-related concerns be fully addressed and patients should receive adequate support to allow them to adjust to and accept devices. Counselling (group or telephone) seems to lead to better outcomes in terms of anxiety, depression and health resource use following ICD implantation (6).
- Congenital heart disease (CHD) refers to problems with the heart's structure that are present at birth. These congenital heart defects, which change the normal flow of blood through the heart, vary greatly in terms of complexity and severity. They range from simple defects with no symptoms to complex defects with severe, life-threatening symptoms(7). The estimated live birth prevalence of congenital heart disease (CHD) in the Republic of Ireland is between 5 and 6 per thousand which is generally reflective of global figures (8).
- Advances in treatment have led to much improved survival rates in congenital heart disease and the transition of children and adolescents to adult care is now a major theme in the literature. Globally high proportions of young people are lost to follow-up and in response there have been repeated calls for formal transition programs which incorporate education throughout adolescence about their condition, its implications, the differences between paediatric and adult services and self-care management (9-12).
- The financial burden of CHD in Ireland is unclear but research from other countries has shown costs can be high and families may have to make sacrifices. Some evidence shows that financial means and material well-being are not as important to quality of life in CHD patients, with more value placed upon family, health, friends and the future.
- Accessing insurance and loans can be challenging regardless of CHD severity, although there is evidence opportunities have improved somewhat. Research on inherited cardiac conditions has shown that the establishment of legislation and guidelines is not sufficient alone and that all players must also be educated.
- Most CHD patients are in employment and work is highly important to them. Those with more complex CHD can experience work-related issues however and for this reason early career guidance is recommended. The majority of ICD recipients seem to return to work especially those who are more educated and have no history of myocardial infarction (13). Some do feel the ICD is an obstacle to their career development but this is not reported by the majority (14) . Very few seem to lose a job due to psychological distress associated with ICD treatment (15).
- In relation to exercise, it is important that those with CHD do participate but also that they do not overestimate their capacity. Symptoms, fears and overprotection by parents and teachers can all act as barriers to participation.
- Although exercise is important, psychological functioning of the patients themselves and their families is a greater determinant of actual quality of life for these patients and can moderate the benefits of interventions such as exercise programmes.

- Complex CHD can lead to more issues in relation to employability and severity of defects seems to be related to cognitive functioning, heart-focused anxiety, parental distress and parental reports of psychological adjustment in their children. Overall, however, research consistently suggests that in patients with congenital heart disease, the severity of their cardiac defect is not proportionally related to quality of life or psychological adjustment (16, 17). Studies have also shown severity to be unrelated to social mobility, insurance access and exercise participation.
- Generally it seems psychosocial adjustment is favourable and similar to healthy peers although a minority of adolescents may be at more risk in relation to decreased psychosocial adjustment or quality of life based on CHD severity, gender, social support and academic performance. There is also some concern that the emotional states of adults with CHD may be artificially inflated in self-report data.
- CHD patients have concerns around contraception, pregnancy and heredity and are not always provided with the information they need. It is currently unknown how those living in Ireland fare in this regard.
- Parents experience a lot of stress at diagnosis and when their child is undergoing surgery and should be provided with support, both social and psychological. Later their support can in turn impact their child's psychosocial functioning. Parental overprotection and delays in shifting responsibility for healthcare during transition to adult care can be an issue.
- Findings in relation to health behaviours in CHD patients are broadly positive for both adolescents and adults.
- There is a clear lack of Irish studies in this area but media reports have suggested peer support groups can be helpful to adolescents in Ireland with CHD, not as an outlet to discuss their CHD but simply to socialise with others who understand their issues, an issue that is reflected in the scientific literature. Feeling different has been established as a feature of CHD throughout childhood, adolescence and adulthood (18-20) and achieving a sense of 'normalcy' seems to be a key process at various stages (20).
- Primary research involving those living with congenital heart disease and their families in Ireland is needed and will be invaluable in assisting the Irish Heart Foundation and the Irish Health services in planning how they can best assist these individuals and their families now and in the future.

Part I: Inherited Cardiac Conditions

Some of the most common inherited heart conditions are listed below:

Inherited heart rhythm disturbances, for example:

- Long QT syndrome (LQTS)
- Brugada syndrome
- Catecholaminergic polymorphic ventricular tachycardia (CPVT)
- Progressive cardiac conduction defect (PCCD)

Cardiomyopathies, for example:

- Hypertrophic cardiomyopathy (HCM)
- Dilated cardiomyopathy (DCM)
- Arrhythmogenic right ventricular cardiomyopathy (ARVC)

Sudden Arrhythmic death syndrome (SADS)

Very high cholesterol levels:

- Familial hypercholesterolemia (FH)

1.1 Inheritable Cardiac Diseases Overall

An Irish study of patients attending a family-based cardiac screening clinic for inheritable cardiac diseases found high levels of anxiety (21). Psychological distress was associated with lower educational attainment, being single or separated and being closely related to the most affected family member (21). This study was cross-sectional but had a high response rate.

A prospective study of persons at risk of a hereditary cardiac disease in the Netherlands, found that emotional distress did not differ from the general population (22). This study involved family members of patients with LQTS, DCM, HCM or ARVC who had received a letter informing them of their increased risk and who subsequently attending the genetic counselling offered. Emotional distress was no greater than the Dutch population average neither before nor after disclosure of their DNA or clinical test results (22).

In another study of inherited cardiovascular diseases (as a group), researchers in the Netherlands found no problems with the well-being of carrier children as compared to representative peers (23). The parents of these children did however rate their child's psychological wellbeing significantly lower. The authors pointed to the lack of differences between carrier children and the reference group as encouraging, providing reassurance regarding the implications of genetically testing carrier children for cardiovascular diseases (23).

The above studies explored carriers and potential carriers of inherited heart conditions. Looking specifically at those with a diagnosed condition as opposed to those at risk or carriers, there does seem to be some impact on patients themselves in terms of health status, anxiety and depression. This impact also seems to vary depending on the inherited cardiac condition in question. For instance a recent study indicated that having a cardiomyopathy (dilated or NCCM) is associated with more averse outcomes in relation to health status, anxiety and depression when compared to familial hypercholesterolemia (3).

1.2 Implantable Cardioverter Defibrillator Recipients

Implantable cardioverter defibrillators (ICD) play an important role in preventing sudden death, a tragic complication of a number of genetic heart diseases (24). A number of studies have explored the experiences of ICD recipients as a group. One review reported that the implantation of the device and associated therapies such as shocks can produce psychological distress (4). Around 30% of ICD patients were reported to suffer from depression and anxiety is also common, with 24-87% experiencing anxiety symptoms following implantation (4). When limited to clinically diagnosed anxiety disorder however, the prevalence ranges instead from 13-38% (4, 25).

Posttraumatic stress disorder can also occur and the review indicated an approximate prevalence of 20% more than twice the estimated general population prevalence (7.8%) (4, 5). Comorbidities and frequent shocks appear to contribute (4). Female gender and longer time to first shock were associated with higher PTSD scores in another study (24) while the experience of sudden cardiac arrest outside of a hospital setting also pushes PTSD prevalence up to rates of 27-38% (26).

Overall, most ICD patients appear to adjust well but some have considerable psychological distress (13). In a study conducted since the above review, anxiety and depression symptoms were related to younger age, living alone and previous history of myocardial infarction or heart failure. Again anxiety was more likely in females and in all patients ICD-related concerns were associated with anxiety, depression and poorer quality of life (13). Overall, the literature seems to underscore the importance of fully addressing patients' ICD-related concerns given the role they seem to play in the level of psychological distress experienced (4). In addition, as the review authors noted, psychological distress in the partners of ICD patients should also be addressed (4).

An earlier review showed ICD patients have favourable return to work rates (62% in largest study included (27)) and suggested the majority of ICD patients who wished to return to work were capable of doing so. Those who return to work seem to be more educated and are less likely to have a history of myocardial infarction (25).

Another study exploring various psychosocial concerns found more than half of patients surveyed were concerned that they had trouble sleeping, difficulty remembering things, were feeling depressed, had overprotective family members or were unable to work (28). Other issues included fear of being shocked, fear that the device would malfunction, dizziness or inability to concentrate. About a third reported a fear of death, sense of loss of control, concerns about having sex or driving a car or inability to engage in hobbies. Finally just 13% expressed concerns about difficulty with interpreting numbers (28).

In 2009, Dunbar et al. found a psychoeducational intervention decreased both anxiety and depression symptoms and health resource use (disability days and calls to providers) in ICD patients in the year after implantation (6). Both group and telephone counselling seem to lead to better psychological outcomes (compared to usual care) following ICD implantation (6).

Some studies have also specifically explored younger ICD recipients. In a study from the Netherlands Koopman et al. found younger ICD patients displayed more anxiety, depression and sleeping disorders (29). Worries were greater among those who had received shocks and those who had had the device for more than 2 years (29). Children with ICDs need help in learning to cope with shocks and should also be monitored for psychological problems (29). An older US study of paediatric patients with ICDs told a more positive story as they found overall psychosocial scores, rates of anxiety and depression and family functioning scores were all the same as normative sample means (30). Patients did seem to experience a greater need for social acceptance however and caregivers perceived their quality of life as lower. Overall, the authors described paediatric patients with ICDs

as resilient (30). As is the case with Congenital Heart Disease more generally (see Part II), severity of disease is not strongly related to quality of life (31, 32). Severity of defect seems to be less important than other issues such as mental health and for paediatric ICD patients quality of life was more strongly related to anxiety, depression and family functioning than severity (30).

In regard to ICD patients, regardless of their age, sufficient support with adjusting to the device and having any device-related concerns addressed seems vital. Poor device adjustment is associated with increased, clinically significant anxiety and depression (33) and some of this distress could perhaps be lessened were more support provided.

1.3 Hypertrophic Cardiomyopathy

Hypertrophic Cardiomyopathy (HCM) is a common cardiomyopathy and affects at least 1 in 500 persons worldwide (34, 35). The disease is associated with heart failure and sudden death. The clinical course of hypertrophic cardiomyopathy is highly variable. Mutation carriers can develop HCM at any point in their life (36). While some patients will remain asymptomatic throughout life, others will develop progressive and ultimately fatal heart failure, or die prematurely due to sudden death (37).

Studies have found quality of life and psychological adjustment to be impaired in HCM patients (38). In a study of mutation carriers rather than patients however, Christiaans et al. found that quality of life and distress were no worse than in the general Dutch population. While psychosocial adjustment seems similar to peers, illness and risk perception related variables do seem to be determinants of quality of life and distress in these individuals and for this reason genetic counselling should address these variables before and after genetic tests take place (38). Another study showed that the majority of mutation carriers value genetic counselling positively and few would prefer not to know. Regrettably however, a substantial portion of carriers are not receiving regular cardiac follow-up and the reasons for this remain unclear (39).

Children diagnosed with HCM are more likely to have no siblings and seem to experience more psychosomatic symptoms (more headaches and abdominal symptoms) pre-diagnosis when compared to healthy children who have a first-degree relative with a HCM diagnosis (35). It has been suggested that these psychosomatic symptoms could be the result of psychological stress due to not being able to keep up with peers in physical activity. The fact that these symptoms disappear after diagnosis seemed to support this theory as children then had an alibi for somewhat decreased exercise tolerance. Children with HCM also seem to become less active after diagnosis (35). Generally, those with HCM are advised to avoid intense physical activity such as competitive sports and to partake in moderate exercise instead (40).

Overall quality of life was not different in children diagnosed with HCM. Self-esteem, peer acceptance and satisfaction with school appear to be normal and family screening for HCM does not seem to negatively influence quality of life (35).

According to a review of Western and Asian literature, individuals with HCM often have high medical costs due to complications, such as treatment for heart failure (including transplantation in severe cases), arrhythmias requiring a permanent pacemaker, ICD implantation, peripheral embolism, endocarditis, fatal and nonfatal stroke and atrial fibrillation (37). Those with hypertrophic cardiomyopathies, especially those with manifest disease, have also reported difficulties in accessing insurance (36).

1.4 Catecholaminergic polymorphic ventricular tachycardia (CPVT)

Long QT, Brugada syndrome and catecholaminergic polymorphic ventricular tachycardia are all inherited channelopathies. Catecholaminergic polymorphic ventricular tachycardia is a rare inherited arrhythmogenic disorder, which can lead to life-threatening ventricular arrhythmias in patients with a structurally normal heart. The age of onset is usually between two and 12 years (41). Patients with CPVT often present with exercise- or emotion induced syncope, but the first presentation can also be sudden cardiac death (1, 42). CPVT diagnosis is often made using exercise electrocardiogram, which typically triggers arrhythmias and treatment consists of beta blockers, often in combination with implantation of a cardioverter-defibrillator (41). Signs and symptoms of the disease usually begin in childhood. Ylanen et al. warn that asymptomatic patients with known CPVT gene defects should also be treated because sudden cardiac death may be the first manifestation of the disease(42). Genetic counselling is recommended and all first degree relatives should be properly evaluated (1). Following a CPVT diagnosis, patients should be counselled on the consequences for themselves and their family. In relation to lifestyle, it is recommended that patients abstain from competitive sports and do not swim unsupervised (43).

Unfortunately CPVT diagnosis is often delayed after first symptoms, which is worrying given treatment with beta-blockers prevents sudden death (44). Any paediatric patients with stress- or emotion-induced syncope must be adequately examined for CPVT as only treatment with beta blockers can prevent sudden death and therefore must be followed for the patient's lifetime (44).

No literature on psychological experiences, social consequences or quality of life in CPVT was found. This seems to represent a significant gap in the literature. This gap may be related to the fact that CPVT is a rare disorder and also due to the fact that the first presentation for CPVT patients can be sudden cardiac death.

1.5 Brugada syndrome

Brugada Syndrome is a hereditary disease which can lead to syncope, and sudden cardiac death from ventricular fibrillation (45).

A multicentre Israeli study of patients with Brugada Syndrome found a high overall complication rate (32%) following ICD therapy and for two of the fifty-nine patients in this study psychological disturbance resulted in the loss of employment (46).

Probst et al. explored the psychological impact of ICD implantation in patients with Brugada syndrome and found quality of life was good in both implanted and non-implanted patients but that implantation does seem to be accompanied by difficulties in patients' social and professional lives (14). The main situations in which implanted patients felt they experienced negative impacts were in purchasing insurance, especially for bank loans, and in their professional careers. These impacts were stronger for those who were young and gainfully employed (14). The Brugada syndrome patients felt restrictions on their ability to develop their career or obtain bank loans due to an ICD were particularly unfair as they judged their health to be good and considered the ICD implantation reassuring (14). The authors agreed restrictions applied by insurance companies and employers seem excessive given the excellent prognosis (very low arrhythmic risk) and almost normal life expectancy of these patients, especially asymptomatic patients. This study highlighted the need to propose specific insurance recommendations to reduce complications experienced by these patients (14).

1.6 Long QT Syndrome

Long QT syndrome is one of the most common cardiac channelopathies and has a high mortality among untreated patients. Those with LQTS are advised to avoid QT-prolonging drugs and high intensity sports (15)

In Norway, a qualitative study explored the psychosocial aspects of living with Long QT syndrome (LQTS), daily life challenges for patients, coping strategies and their experiences with healthcare services (47). Early and gradually acquired knowledge of the syndrome was reported to be an advantage. Patients reported experiencing worries and limitations in daily life but main concerns were for their children or grandchildren. They found healthcare providers knowledge of LQTS to be minimal and this resulted in uncertainty, misinformation and worryingly, even wrong advice regarding treatment (47).

A Dutch study found predictive testing for long QT syndrome does lead to distress in carriers and their partners in the short-term but that these distress levels return to normal within 18 months (48).

Parents are also affected and those with carrier children report greater distress. These increased distress levels are prolonged and were clinically relevant in some cases. Parents were particularly concerned about possible hazardous behaviours during puberty (49). Mothers of children with diagnosed LQTS also appear to have higher anxiety compared to other mothers (50).

In a US study, parents also described fear of their children dying and frustrations about lack of LQTS knowledge among healthcare providers (51). The authors also noted that the impact on the lives of both children and their families seems to be more significant when an LQTS diagnosis is established during adolescence (51).

Children with LQTS seem to suffer with anxiety and fear of failure and criticism. They have significantly more internalising problems and tend to keep feelings to themselves and minimize their feelings of anxiety rather than share their feelings openly (50). Mothers of these children also seem to have higher anxiety than other mothers (50).

In a paper on genetic discrimination and health insurance, Hudson et al. described a case where a healthy active 4-year old was twice denied health insurance (52). This child's mother had died in her sleep of sudden cardiac arrest and his uncle had also died of sudden cardiac arrest in his twenties. These sudden and unexpected deaths led to genetic tests which revealed this child had inherited the altered gene for long QT syndrome from his mother and as result of this the child's father has had difficulties securing family health insurance which will cover his son (52).

1.7 HCM and LQTS

A number of studies group HCM and LQTS patients. In Norway, patients with familial LQTS or HCM had higher levels of general anxiety and their avoidance or fear was independently related to their general anxiety, depression and physical health. The fact that avoidance and fear are potentially modifiable and might be important determinants of general anxiety, depression and physical health in these patients provides support for the further development of genetic counselling. The researchers also found that a recent sudden cardiac death in the family was independently related to general anxiety and depression regardless of disease status (53).

In a qualitative study of patient experiences in the UK, patients who had undergone testing for HCM or LQTS were interviewed. The majority embraced testing and screening for themselves and their children but concerns arose in relation to communicating about these disorders, deciding whether to involve elderly relatives and pressures relating to family responsibility (54). There was also some

ambivalence about the value and impact of DNA test results and this may influence disposition towards testing and issues around family communication (54). Reactions to positive test results varied from outwardly unconcerned, as a positive result was expected, to guilt and anxiety towards other family members. Negative results were met with relief but also shock and distress regarding the unnecessary impact their now overturned diagnosis had had on life up till now (54).

1.8 Dilated Cardiomyopathy

Dilated cardiomyopathy (DCM) is the most common form of cardiomyopathy accounting for one third of cases (55). Roughly 750,000 people in the United States have dilated cardiomyopathy and approximately half of these cases are familial (56).

Patients with DCM have reported pronounced restrictions in quality of life and psychological wellbeing including significantly impaired physical functioning and role limitations due to: physical and emotional problems, social functioning, mental health, perceptions of general health, sleep and vitality. They have also displayed anxiety and depression levels higher than general population samples (57). Interestingly, those with familial cardiomyopathy seem to display less impairment in quality of life when compared to non-familial cases (57). Researchers have suggested that patients with DCM may benefit from efforts to improve psychological adjustment to their condition (57). In an Italian study, DCM patients who showed significant reverse remodelling in the first two years of treatment showed survival comparable to the general population (58) and should therefore as the authors argued be considered for life insurance coverage, at least for short or medium term coverage. This study showed survival probability does strongly depend on individual treatment and evolution of the disease and is measurable within the first two-years of follow-up meaning insurance companies could use this data to improve their risk stratification (58).

1.9 Arrhythmogenic Right Ventricular Cardiomyopathy

Patients with arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) specifically appear to have elevated body image concerns and device-related distress, especially among younger patients (33). These patients showed elevated levels of general clinical anxiety and ICD-specific anxiety and younger patients in particular appear to struggle with device acceptance (33).

1.10 Familial Hypercholesterolemia

Familial hypercholesterolemia (FH) is a common inherited metabolic disease with a prevalence of 1 in 500 in most Western countries (59).

A qualitative study of patients with FH found their personal sense of vulnerability to cardiac disease can shift dramatically with changes in situational factors such as cardiac events in the family, illness experiences or becoming a parent (60).

Children treated for FH do not appear to have greater psychosocial dysfunction than their peers. They have shown similar psychosocial scores and the prevalence of psychiatric disorders is not increased (61). The exception was children who had lost a parent or had a parent who had cardiovascular disease due to FH, these children did have higher symptom scores in relation to school and expression of anger (61).

Similarly, in the Netherlands, quality of life in those screened for FH remained unchanged during screening and there were no differences between FH positive and FH negative subjects in this regard (62). The authors took their finding of no adverse effects on short or long term quality of life in this longitudinal study as evidence that the set-up of the current screening programme is adequate(62). In the Netherlands, Marang-van de Mheen et al. showed that in spite of existing guidelines and legislation on the use of genetic test information, screening participants still encounter

unanticipated insurance problems (59). It is unclear whether these problems are the result of insurance companies asking questions or individuals giving more information than asked for but the conclusion of the authors was that guidelines and legislation on genetic information alone are not sufficient and the education of all players involved is of equal importance (59).

Part II: Congenital Heart Disease

2.1 Incidence of Congenital Heart Disease in Ireland

The estimated live birth prevalence of congenital heart disease (CHD) in the Republic of Ireland is between 5 and 6 per thousand which is reflective of overall incidence in terms of magnitude (8). Overall incidence as per studies of various populations throughout the world is 8 per 1,000 live births with incidence of defects requiring surgery or intervention at 3.5-4/1000 live births (63). While incidence in general reflects global figures, incidence of major congenital heart disease (MCHD) (including hypoplastic left and right heart, coarctation and pulmonary arteria) is said to be high in Ireland. In 2009, among 75,720 live births in Ireland, 115 infants were born with MCHD (8). This study represents one of the very few studies relating to congenital heart disease in Ireland. Just 44 (or 21%) of these diagnoses were antenatal, a figure which compares poorly with Europe (8).

2.2 Increased survival

Developments in the management of CHD have led to significantly improved survival rates for many CHD diagnoses (64). Since the advent of neonatal repair of complex lesions in the 1970s, approximately 85% of patients survive into adult life (12). A more recent Belgian study suggests that almost 90% now have the prospect of surviving into adulthood (32). These improved survival rates have in turn created the need for effective transition of CHD patients from paediatric to adult care.

2.3 Transition

As with many chronic conditions (31), the transition from paediatric to adult care in Congenital Heart Disease (CHD) is both complex to manage and key to continuity of quality care. Young adults with CHD are at high risk for complications in later adulthood and thus must be made aware of the need for regular medical follow-up (11). The need to develop programs that assist adolescents with congenital heart disease and their families in dealing with special healthcare needs, allowing them to successfully move into the adult world, has been described (65). A range of transitional health-related issues for adolescents with CHD have been identified, including medical follow-up but also insurability, employability, sexuality and reproduction (11, 65). Some of these additional issues will be discussed at greater length in later sections of this review.

Successful transition into the adult healthcare system requires early preparation and counselling of patients with CHD, well before they turn 18 (66). The American College of Cardiology Task Force have recommended a formal transition process be used in order to provide optimal transfer of patients into Adult Congenital Heart Disease (ACHD) care (12). This report highlighted the difficulty of transitioning from childhood to adulthood and how this difficulty increases where residual cognitive and physical disabilities are present (66, 67). Specifically, they recommended that this process should begin at 12 years of age and be tailored on the basis of the patient's individual maturity level (66). Both physical and emotional maturity are required and the age at which this occurs varies depending on the patient. Strategies for patient transfer should be based on a stepwise approach towards establishing autonomy and understanding one's cardiac problem and the lifestyle issues pertinent to long-term stability of CHD (12, 68). These recommendations have since been echoed by the American Heart Association who advised timing of transition be guided by emotional maturity and developmental level and that adolescents should be engaged in their own transition planning (17).

During transition both the patient and the healthcare provider must change their approach to medical care and both require knowledge and skills to make this change. The process has been described as demanding a shift from paediatric family-centred care to adult patient-centred care (69). Patients must learn strategies for managing health, how to cope with medical decision-making and face morbidity and possible early mortality (69). In the US, a web-based module has been shown

to be a successful tool in the education of paediatric residents about the importance of transition and the recommended guidelines regarding transition (70).

Gurvitz and Saidi also highlighted the importance of educational and vocational transition in preparing patients for a productive adulthood (69). This echoes the earlier recommendation of the American Heart Association who called for early education on later insurance, employment and health (17), citing the positive effects revealed by long-term studies (71, 72).

In Canada, researchers interviewed CHD patients, their parents and healthcare providers about transition and found a lack of clear role expectations. Less than 40% of the patients had a clear understanding of transition and its implications for their cardiac care (11). Findings from an earlier study suggest that while adolescents do recognize the need to take an increasingly active role in managing their own healthcare, they are unsure how to begin (73).

Children with more knowledge about their diagnosis demonstrate a better understanding and are more likely to communicate directly with health care providers (11). Reflecting on their findings, Clarizia et al., echoing US researchers, again called for a formalized, systematic approach to transition preparation which emphasizes comprehensive education (11).

2.3.1 Transition in Ireland

A study on transition for CHD patients in Ireland has yet to be conducted. Anecdotal information comes in the form of an Irish Times article where a nurse discusses adolescents with CHD transitioning from Crumlin hospital's paediatric service after the age of 16 and how she manages this process with them (74). She highlights the increased demand for adult services. The Adult Congenital Heart Service, which is based in Dublin's Mater Hospital, began just over 10 years ago. Initially they were receiving 220 new referrals per year but this has since more than doubled and it is estimated that about 90% of these new referrals are children "transitioning" from Crumlin's paediatric service after the age of 16 (74). The nurse specialist interviewed highlighted that transfer to adult services is often more difficult for parents. During the first visit, she reports she 'makes sure they understand their condition, their medication, what surgery they've had and which symptoms they should be concerned about and report in between visits to the clinic' (74). A formal scientific study into this issue is vital to inform current services in Ireland.

2.3.2 Consequences of Inadequate Transition

Failing to properly address transition of patients with CHD can lead to loss of health care coverage and inadequate continuity of care (67). The American Heart Association warns that the absence of structured programs to guide transition can lead to delayed or inappropriate care, improper timing of transfer of care, and undue emotional and financial stress on patients, families and the healthcare system (17).

A study of the long-term outcomes of CHD patients who underwent paediatric surgery in Finland revealed more than half of patients who required cardiac follow-up had dropped out (75). Most of the patients who had dropped out believed they were in good condition (75). Unfortunately lack of adequate cardiac follow-up is a frequent issue in this patient group and has been documented in several studies (76-78). Most recently, a systematic review revealed high proportions of young people are lost to follow-up or experience long gaps in care after leaving paediatric cardiology (9). Nieminen et al. argued that unsuccessful transfer of care is the likely cause for this high rate of follow-up dropouts. In order to address this, they called for an intense effort to develop an efficient chain of services to ensure the effective transition of patients and expertise from paediatric to adult cardiac follow-up (75). If non-attendance is an issue in Ireland, qualitative research could be very useful in exploring the reasons why patients who need to are not attending regularly.

2.4 Financial Burden of Illness

The improvements in survival rates (12, 32, 64) in CHD have led to lifelong interaction with health services for many people, resulting in increased costs for parents and society, and later patients themselves. Some European research suggests that the financial cost of CHD treatment and management may be high (e.g., Czech Republic data) (79) and expenses can be considerably higher than those for general population counterparts, with a large portion of costs being paid directly by parents (e.g., in the Netherlands)(80). Research conducted in Pakistan indicated potentially significant negative financial implications for families of children with congenital heart disease, including having to take unpaid leave, losing jobs or business and selling assets (81).

While financial burden seems a very real issue in CHD (79, 80, 82), a study on individual quality of life in adults with CHD revealed financial means and material well-being were not high on CHD patients' list of concerns (83). This cross-sectional, comparative study was conducted in Belgium and involved 579 CHD patients and 446 matched controls. Compared to control subjects, significantly fewer patients indicated financial means and material well-being to be important for their quality of life. Moreover, the relative importance of these domains was also significantly lower for CHD patients (83). Family was the most significant determinant of quality of life in adults with CHD, followed by health, friends and future. Environment, financial means and material well-being, and nourishment were of less importance. In attempting to explain these findings, the authors suggested that severe heart conditions or previous operations may lead patients to put material aspects in a different, less valued, perspective.

2.5 Insurability, Mortgage and Loan Applications

Life and health insurance availability vary greatly both within and between countries. In general, insurance for adults with CHD is often difficult or expensive to obtain and this restricted access can be an added stressor (84). It is worth noting that while this review did include European studies, studies in this area seemed more focused on the US system of insurance through employer which is quite different to the current system in Ireland.

In the US, access to comprehensive health care coverage for congenital heart disease patients is often limited due to their 'pre-existing condition' (85, 86). In the case of congenital lesions with higher mortality ratings life insurance also can be difficult to obtain (85). The Second Natural History Study of Congenital Heart Defects included questions related to insurability and employability. Gersony et al. reported just 16% had no health insurance, while 35.7% had no life insurance (76). More recently, Sable et al. also reported that a significant proportion of adult CHD patients in the US have difficulty obtaining and/or continuing health insurance (17).

Over the last few decades, insurance opportunities are said to have improved for CHD patients. While historically adult patients found it difficult to qualify for life and health insurance, this situation appears to have improved and now many adults who have undergone successful surgical repair may be eligible for both health and life insurance at standard or higher rates (65). Nonetheless, in Switzerland, where health insurance is obligatory, a significant portion of adult CHD patients (21%) still report problems with insurance in general, and with health insurance in particular (67%) (87).

In addition to insurance, mortgage and loan applications can also present a challenge. In the UK adults with CHD are more likely to have difficulty obtaining life insurance or a mortgage regardless of whether CHD is mild, significant or complex (88). Similarly in France adults with CHD are considerably more likely to have difficulty obtaining a mortgage or loan and this difficulty is again

independent of disease severity (89). Condition severity has an impact on whether insurance companies require payment of surplus fees, with insurance companies asking significant and complex CHD patients to pay surplus fees. Age, sex, other diseases, cardiovascular risk factors and duration of the loan were not predictors of loan application outcome and less than 20% did not report their CHD (89).

2.6 Employment

2.6.1 Employment rates

Research on employment rates for those with congenital heart disease generally yields positive findings. Studies in the Netherlands showed that, while adults with CHD generally have lower educational and occupational achievement - even those with milder defects (90, 91), the vast majority (78%) of these adults were still living independently and showed favourable outcomes regarding marital status (90). In a Finnish study of long-term outcomes, the overall employment rate among those who had undergone paediatric cardiac surgery actually exceeded that of the general population (75). Notably, the employment rate for the subgroup of patients with cyanotic defects was lower than the general population rate but this difference disappeared once the researchers controlled/accounted for intellectual disability (75). Studies in Sweden (77%)(78), the UK (approximately 66% excluding students, those on maternity leave and those who had retired)(92) and Belgium (almost 60%)(83) have also revealed that the majority of CHD patients are in employment. In a US study involving patients with aortic stenosis, pulmonary stenosis, or ventricular septal defect, unemployment rates were similar to national averages except for women with aortic stenosis, who had a significantly higher unemployment rate than did age- and sex-matched controls (76).

Findings in relation to complex CHD are in general less encouraging. In the Netherlands, patients with complex CHD had reduced job participation compared with patients with mild CHD and the general population. For instance, 64% of patients older than 25 years with complex CHD were employed compared to 83% in the general population (93). In Sweden, Ternstedt et al. also found those with mild defects had better employment status (94) and in the UK more than half of patients with complex CHD experience problems in their careers (92).

More complex forms of CHD such as atresia and hypoplasia of a valve or chamber and ventricular inversion require regular cardiac follow-up and patients may have limited exercise tolerance and are sometimes unable to maintain full employment (84). Patients may not have reached typical educational milestones or developed employment skills due to real or perceived restrictions and/or fear of limited lifespan. Both physical limitations and lower educational attainment can then restrict employment opportunities (84). These limitations can in turn deny patients the ability to support themselves independently (84). Indeed research shows that adults with CHD are more likely to maintain independent lifestyles and live with their parents versus healthy peers (95).

Many complex CHD patients therefore receive disability benefits or experience career problems or job handicaps. They tend to cease employment due to physical problems/demands, emotional problems, or problems with transportation. The most frequent requests for adaptations to the work environment focus on more flexible work hours, reduced time pressure or workload, and increased freedom to organize one's work (17, 92, 93).

Overall, it seems that most adults with congenital heart disease experience similar employment patterns and issues to the general population. Those with more complex CHD, however, do tend to face issues when it comes to employment.

2.6.2 Early career guidance advice

The most straightforward solution to improve the employment status of CHD patients is helpful, positive, medically appropriate advice regarding potential careers (96). In the UK, Crossland et al. found that receiving career counselling was significantly associated with increased employment in CHD patients (92). However, just one-fifth of CHD patients in this study had received counselling regarding their career (92).

The need to attend to social as well as medical aspects of care has been highlighted (65). Career and vocational counselling have been identified as an integral part of any adolescent CHD program (65, 93). Price has also recommended early discussions may help individuals with congenital heart disease to develop appropriate expectations and informed career choices (97). Research repeatedly recommends that adolescents with CHD be assisted in selecting a career that reflects their personal interests and level of education as well as physical and emotional capabilities (17, 65, 86, 93). Where career counselling is provided to CHD patients the source should be both qualified and knowledgeable. Advice is often provided by medical staff not trained in this field or career advisors not trained to give advice based on the medical condition of the patient (96).

In relation to employment discrimination, in the US and France legislation has been enacted to reduce employment discrimination of individuals with pre-existing disabilities and legislation now supports many workers once they can carry out the job for which they were hired (65, 86, 96). It is unclear yet whether there is a need for the same in Ireland. Perhaps interviewing patients here will shed some light on issues they may be facing and whether supportive legislation would be helpful.

2.6.3 Importance of employment

Difficulty securing employment and insurance can be sources of frustration for CHD patients (84). Nieminen et al. point to the high employment rates among CHD patients in Finland as evidence of the high value of work among this patient population. Indeed, the study of adults with CHD by Horner et al., in Australia, found the majority were highly committed and functioning well with regard to both education and employment (18). Work was highly valued and, in general, people with congenital heart disease showed enormous resilience in continuing to work no matter how constricting their health status. These patients worked in a wide range of occupations and tended to keep their CHD a secret from peers and employers (18). Employment may be a protective barrier against fears of decline and premature death, and feelings of loneliness, isolation, anxiety, and depression. As Horner et al. found individuals who were single, unemployed and isolated were more disabled by these feelings (18). Lack of employment is also associated with poorer quality of life (84, 98) and in a study of Czech patients there was a positive association between depression and unemployment (99).

2.6.4 Social mobility

In relation to social mobility among CHD patients, a German study found that once patients enter the labour market intergenerational social mobility rates are comparable with the general population. Social background is a key determinant of upward mobility and for patients who do enter the labour market CHD severity is not a determinant of social mobility (100).

2.7 Exercise

2.7.1 Subjective experience

Chiang et al. interviewed adolescents with mild CHD to understand their subjective experience of exercise. They found that when these teenagers faced and accepted their own exercise limitations, they would assess their condition and develop coping strategies based on their accumulated

experiences and participate in exercise to maintain a healthy body and mind and live a normal life (101).

2.7.2 Peer comparisons

In Northern Ireland, Casey et al. found the majority of 4-5 year old CHD children had good exercise tolerance but maternal worry does influence activity levels (102). In a study of adolescents with mild CHD, Chen et al. found approximately half did not follow the exercise intensity recommended by cardiologists. They recommended exercise instruction interventions noting that inadequate exercise patterns can lead to cardiovascular complications (103).

Young adult patients with a history of CHD surgery show diminished exercise capacity, physical activity and quality of life in comparison to healthy peers (104). Exercise intolerance is prevalent (105) and generally, except for very young men who report normal health, young adults with CHD describe poor health and exercise limitations relative to their peers in the community (106). Patients who do not exercise enough have reported desire to participate in exercise (107).

2.7.3 Barriers

The most common barriers to exercise according to Swan et al. are symptoms and fears (108). Other studies have shown overprotective parents and teachers and SES can also contribute to activity levels and exercise tolerance (102, 105, 109).

A study by Prapavessis et al. suggests that CHD patients' beliefs regarding physical activity are not a limiting factor in engagement in more exercise. It seems that perceived group norms and perceptions about how much control one has over one's own behaviour are more predictive of exercise intention and exercise behaviour (110). Another study found that among teens self-efficacy was more important than CHD severity in determining exercise participation (111). Healthcare professionals may play an important role in addressing exercise barriers given cardiologist recommendations have been shown to significantly influence both parental attitudes and self-efficacy (111).

2.7.4 Safety

Naturally there are safety concerns in relation to CHD patients and exercise. Cardiopulmonary stress testing has been recommended for children who have had congenital heart surgery (112). This procedure provides additional information and thereby allows more objective recommendations regarding the most suitable sports and right levels of intensity (112). In Ireland, a specialist nurse working with adolescents with CHD reports exercise recommendations are tailored to the individual (74). A systematic review found that physical exercise training programmes for children and young adults with CHD do seem to be safe and improve fitness. Indeed, none of the studies reported negative findings related to training (although cardiac effects have not yet been studied to any extent). It is therefore recommended that CHD patients participate in physical exercise training (113).

2.7.5 Overestimating exercise capacity

Many adults with CHD do not meet normative expectations for perceived health status and exercise tolerance (106); however some can overestimate their capacity. In CHD research, exercise is often looked at in combination with quality of life and researchers recommended exercise tests and quality of life instruments should be used together to get an appropriate overview of the health status of patients with CHD (114). In comparing self-reported quality of life and exercise capacity, researchers found a substantial number of patients actually overestimate their physical capabilities (114). For example, in one study 44% of patients thought all exercise was safe (108) and another study found adolescents and adults significantly overestimated actual exercise capacity based on

exercise test results (115). This may also be the case in Ireland given the anecdotes regarding young men and weightlifting reported by the nurse interviewed by the Irish Times (74).

2.7.6 Benefits

Dulfer et al. found that participation in an exercise program improved health-related quality of life in children (aged 10-15) with CHD (specifically tetralogy of Fallot or a Fontan circulation) although there was no change for those aged 16-25 (116). Participating in the exercise group significantly improved self-reported cognitive functioning and parent-reported social functioning (116). A study by Morrison et al. showed exercise training in adolescents with CHD was safe, feasible and beneficial. No adverse effects or mortalities were reported and it improved activity (117).

Muller et al. also called for the promotion of an active lifestyle after finding most adults with CHD are fairly active and daily physical activity is positively correlated with exercise capacity (118). In their study however there was no association between physical activity and quality of life (118). In fact, as the same researchers discovered in another study, depression (even minor symptoms) seems to have a larger impact on quality of life than exercise capacity (119). The benefits of exercise participation in terms of health-related quality of life also seem to be moderated by the mental health of parents. In a study of exercise training for adolescents with CHD, Dulfer et al. found adolescents whose parents had poorer overall mental health, anxiety, insomnia, or severe depression reported a decrease in social functioning (120). Thus, it seems that - while exercise is important - psychological functioning of the patients themselves and their families is a greater determinant of quality of life for individuals with congenital heart disease.

2.8 Family Socioeconomic Status

While some studies have examined socioeconomic status and prevalence of congenital heart disease, there is little literature on the importance of family socioeconomic status in relation to congenital heart disease and quality of life. One study conducted in Bosnia and Herzegovina found that when parents were interviewed, low socioeconomic status seemed to influence various aspects of health-related quality of life in children with CHD. These included physical health and activity, school activities, psychosocial and emotional health and social activities (121). In contrast to their parents however, the reports of the children with CHD themselves showed no statistically significant difference in their health-related quality of life based on family socioeconomic status (121).

In Northern Ireland, in a study of 4 and 5 year old children after intervention for congenital cardiac disease found that higher levels of social deprivation were associated with more GP and A&E visits (102). In a study conducted in England, lower socioeconomic status was related to poorer exercise capacity (105). This association was strongest in patients with mild defects living in the most socioeconomically disadvantaged communities. Those with lower socioeconomic status were also more likely to smoke and have diabetes (105).

2.9 Romantic relationships

Studies by Kokkonen and Paavilainen (95) and Gersony et al. (76) found that people with congenital heart disease were less likely to be married, or married at a later age. Winter et al. (122) also reported that there were fewer CHD patients in relationships but found that the relationships were more satisfactory. More positive findings come from long-term studies. For instance, Van Rijen et al. (90) reported that the majority of CHD patients (at least 72%) were in some sort of relationship (40% were married, 11% stable relationship) while Nieminen revealed that CHD patients were living in a steady relationship as often as the general population (75).

2.10 Sexual functioning

Impaired sexual functioning occurs in both males and females with CHD (123). A study in Germany found that males under 40 with CHD engage in sexual relationships less often. Patients report fears before or during intercourse and physical symptoms such as dyspnoea, arrhythmia or chest pain. Furthermore a prevalence of 10% was found for erectile dysfunction and this was strongly associated with subjective wellbeing, especially depression (prevalence of 3.2% up to 33.3% with ED) (124). Cook et al. (125) found a higher prevalence of erectile dysfunction in men with CHD (38%), which was strongly related to use of beta-blockers but unrelated to CHD complexity. Most men attributed symptoms to their CHD but also reported they were willing to discuss these issues with their cardiologist (125).

Research findings on satisfaction with sexual functioning in congenital heart disease are somewhat inconsistent. Norwegian researchers found that fear is associated with sexual dissatisfaction in males with CHD, while for women issues include fear of pregnancy, rejecting partner's sexual initiative and conflicts relating to sex (126).

Other studies have found sexual satisfaction equal to the general population (122), with a minority (10-20%) of CHD patients reporting problems with sexual satisfaction (127). It seems that satisfaction with sexual function is not always an issue for CHD patients. However, sexuality is an important aspect of quality of life for these patients (122) and as a result where problems do occur they can be the source of much distress (127). While partners remain relatively unaffected, patients perceive lower body esteem, decreased sexual esteem and more distress during sex (122). Patients are not fully informed and thus worried about use of contraceptives, heredity, pregnancy and delivery. To counteract this, it is important that CHD patients receive appropriate education in sexuality, pregnancy, delivery and hereditary issues from a young age (123), with doctors receptive to discussing sexuality issues with their patients and to providing patients with access to adequate therapy where necessary.

2.11 Contraception, Pregnancy and Heredity

Thanks to the major advances in diagnostic and surgical methods, people with congenital heart disease (CHD) now survive into and beyond their reproductive years thus bringing issues of fertility, contraception, and childbearing to the fore (128-130). Both contraception and pregnancy can be associated with increased risks for women with CHD (131). Oestrogen contraceptive pills are contraindicated for those with cyanotic CHD or residual pulmonary hypertension and intrauterine devices increase risk of endocarditis (84). For many women with mild cardiac defects, pregnancy is generally well tolerated with no long-term consequences. However, for those with more complex defects, pregnancy, labour and delivery require closer monitoring and may in some cases be contraindicated (84). As the US Centers for Disease Control and Prevention advise, women with CHD may need to undergo procedures prior to becoming pregnant or take certain medications during pregnancy. In addition offspring may be at risk for having a heart defect and therefore seeing a genetic counsellor may be helpful (132).

In a study conducted in Canada, Kovacs et al. found that many women with CHD lack adequate knowledge regarding contraception and pregnancy risks (133). This echoed the findings of an earlier study by Belgian researchers, who found that only a quarter of patients knew correctly whether or not, and to what degree, their congenital heart disease could be inherited by their children (134). While 80% of female patients knew that the contraceptive pill was likely to be the most appropriate contraceptive, only 27% knew whether or not an intrauterine device was contraindicated. Moreover, most women (87%) were not aware of the complications of pregnancy (134).

In the Netherlands, in a study which included males, the majority of patients reported disease specific worries and fears regarding the use of contraceptives, heredity, pregnancy and delivery (123).

Contraception, pregnancy, and genetic counselling are issues of great concern for adults with congenital heart disease (135, 136). In the US, a study of 124 people with congenital heart disease found that 22.8%, 28.2% and 15.7% had concerns about contraception, pregnancy and genetic counselling respectively. Of those who had concerns, less than half (22.2%, 36.4%, and 47.4%, respectively) did not have their questions answered in full and to their satisfaction (136).

As women with CHD who become pregnant may experience anxiety associated with increased risk or recurrence of CHD in their offspring (3-14%), it has been recommended that this be addressed early in the second trimester with foetal echocardiography which can now screen for significant foetal CHD (84). Women with complex CHD appear to have the highest levels of concern regarding their fertility and risk of genetic transmission of CHD, as well as concerns about adverse effects of pregnancy on their own health. It is recommended particular attention be given to discussing sexual health with these women (137).

It is important to establish whether individuals with CHD in Ireland are currently receiving sufficient information and advice on issues around pregnancy, contraception and heredity. In particular, it seems worth investigating whether women with complex CHD feel they are receiving adequate support.

In relation to parenthood, Gersony et al. also found elevated childlessness among women with CHD (76) whereas Van Rijen et al.'s study, which included males, found more favourable outcomes in relation to offspring (90). The Finnish study of long-term outcomes found the number of parents among patients was just slightly lower than the general population (47% vs 49%, respectively) (75).

2.12 CHD with Down Syndrome

The incidence of CHD in Down's syndrome is high at approximately 43% (138). The American Academy of Pediatrics (AAP) Committee on Genetics recommend infants with Down syndrome receive an echocardiogram and an evaluation from a paediatric cardiologist soon after birth (139). In Ireland guidelines recommend a high level of clinical suspicion of congenital heart disease for all new-borns with the syndrome, examinations and further examinations later if any signs or symptoms develop (140). Further exploration of this specific population is beyond the scope of this review.

2.13 Parents/Family

2.13.1 Diagnosis, Surgery, childhood and familial issues

Increased stress at time of diagnosis occurs regardless of whether diagnosis is made pre or post-natally, though Brosig et al. showed that 6 months after birth stress levels in parents of those diagnosed postnatally had returned to normal whereas those who received prenatal diagnosis still had significantly higher stress levels. More severe defects can also lead to greater parental distress (141). Parents should be provided with psychological support whenever diagnosis occurs (141) and those diagnosed prenatally may require counselling throughout pregnancy to help them recover from the loss of the imagined healthy child (142).

The demands of care for infants with congenital heart disease are a source of stress (143) and hospitalization of children for CHD surgery is also known to be a stressful experience for parents. In a UK study Franck et al., found little difference in stress levels between mothers and fathers but did find stress was higher for those from more deprived communities and those born outside the UK

(144). Some studies suggest mothers experience greater distress than fathers but fathers of children with CHD are more distressed than fathers of children with other diseases (82). As is the case in parents of children with other diseases, factors such as employment status and financial situation influence distress and hopelessness in parents (82, 145). The increased emotional distress experienced in relation to surgery often resolves after 12 months but mothers of children with CHD often continue to feel that they are not in control and life events are largely a function of fate (146).

Clearly having a child with CHD affects psychological functioning in parents and is associated with stress. Moreover, parents can also affect the functioning of the child with CHD. For example, Casey et al. found the majority of 4-5 year old CHD children had few residual symptoms and good exercise tolerance but maternal worry negatively influenced activity levels (102). Maternal worry was also a predictor of frequency of unscheduled health service demands and seems to lead to more GP and A&E visits (102).

In a grounded theory study looking at parenting young children with CHD, a process of 'Parenting under Pressure' emerged and this was characterised by four overlapping and re-emerging phases: "(1) realizing and adjusting to the inconceivable; (2) growing increasingly attached; (3) watching for and accommodating the unexpected; and (4) encountering new challenges" (147). The importance of social support for parents has also been highlighted, as anxiety and depression seem to be more prevalent in parents lacking social support before their child's surgery (148). Souvie et al. reviewed 25 studies and recommended clarification of healthcare professionals' roles in order to ensure provision of psychological support to parents and that parents should also be referred to community-based ongoing social support services (149).

2.13.2 Adolescence and beyond: Support helps but not overprotection

The psychosocial functioning of parents has also been found to have important effects in the adolescence and adulthood of CHD patients. Anecdotally a nurse in Ireland has commented on transition being harder on parents and one mother compared her son's nonchalant attitude to her experience of 'still being traumatised' (74). However, a scientific investigation would provide better evidence of any such issues.

Paternal support can help in relation to the psychosocial functioning of adolescent children with CHD (decreased depressive symptoms and loneliness) (150) but in adult patients perceived parental overprotection is associated with heart-focused anxiety (151). It should be noted that, unrelated to parental influence, patients can also over-perceive heart problems themselves, particularly if they also suffer from anxiety (152). Furthermore patients own poor illness perceptions (beliefs about their illness) independently predict their quality of life (153).

Many adults with CHD recall parental overprotection during their teenage years (17, 154) and this parental anxiety and overprotection may explain parents' reluctance to shift more responsibility for illness management to the adolescent (17). A study in Canada showed parents continuing to be highly involved in care activities during adolescence, with 95% accompanying their child to visits at the clinic, 68% staying with their child for the entire visit and 45% administering their medication (11). As discussed in the transition section it is important for the adolescent to take an increasingly active role in managing their own healthcare. It seems, for parents, there is a balance to be achieved in terms of support without overprotection and allowing adolescents to transition into adult care by gradually taking more responsibility in relation to their own care.

As in childhood, it is important parents receive adequate support themselves. This will also contribute positively to the functioning of their adolescent with CHD. We know for instance parental mental health moderates the benefits of exercise participation and adolescents whose parents have

poorer overall mental health, anxiety, insomnia, or severe depression can experience a decrease in social functioning (120).

2.14 Growing-up with Congenital Heart Disease: Summarising the various life stages

2.14.1 Childhood

In childhood, congenital heart disease (CHD) can hinder friendships and make children feel different or not normal. Children with complex CHD face issues such as extended absences from school, cyanosis, scars and sports restrictions. These issues in turn hinder friendships and conflict with a sense of “normalcy” leading children to feel different (18). Gender differences exist in that teasing and low self-esteem occur more often in boys (18).

In the Netherlands, children who have recently undergone invasive treatment for CHD show poorer intellectual functioning in several areas including verbal IQ, verbal comprehension and perceptual organisation. Overall though they have IQ scores within the normal range and school-related behavioural and emotional adjustment is favourable and similar to peers (155).

A recent study found children with CHDs receive special education services more often than children without birth defects. These findings were said to highlight the need for special education services and the importance of developmental screening for all children with CHDs (156). This echoes the recommendations of Spijkerboer et al. who further explained the importance of remedial services (155). Problems at school combined with reduced physical ability could result in reduced self-esteem in children with CHD. Removing or stabilizing delay via special education services can help children to cope at school and thereby potentially reduce the risk of development of psychopathology (155).

This psychopathological development in children with CHD can emerge in the form of behavioural disorders, both externalising (e.g. aggression, hyperactivity) and internalising (depression, anxiety) (157). Much of the research exploring psychological adjustment, quality of life and developmental problems in CHD groups children and adolescents together. Findings in relation to these issues are discussed in greater detail below (see section 16.2.2).

Psychological intervention has also been shown to be effective in promoting adjustment in children with CHD entering school and their families. In the UK, a randomised controlled trial of psychological interventions to promote adjustment in children with congenital heart disease and their families showed no difference in child behavioural outcomes but improvements in maternal mental health and family functioning, and fewer school days missed and children perceived as ‘sick’ less often by their mother (158).

2.14.2 Adolescence

In Australia, Horner et al. interviewed adult CHD patients and asked them to recall their childhood and adolescence. They identified themselves as normal teens who did not identify as ill or disabled. Complex CHD did not seem to interfere with education and those who did suffer health complications displayed great resilience and determination to complete their education (18). These results are supported by Nieminen et al.’s study of long-term results, in which the overall educational profile of CHD patients was comparable to that of the general Finnish population (75). Most had more than compulsory education and 10% had a university degree. Education levels were lower among patients with an atrial septal defect or cyanotic defect (75).

Generally adolescents with CHD reported normal dating though for males with cyanosis, dating experiences to be minimal and many adolescent CHD patients were self-conscious in relation to their

bodies and scars. Females reported employing tools such as makeup, nail polish and clothing to hide their CHD (18).

Overall, the findings of Horner et al.'s study suggested that adolescence is a relatively healthy time for most and more enjoyable than elementary school (18). It is worth noting however that in this study, adults were interviewed and asked to recall the impact of CHD now and at earlier stages of life. Thus these reflections on adolescence are based on recall and not ongoing experiences.

In another study, current adolescents and young adults in the US did report feeling different and set apart from their peers (73). Common dilemmas and concerns included whether or not to disclose their CHD to others, fear of rejection and other social consequences and fear of forming close relationships (73). There was also concern about planning for the future due to their uncertain longevity (73). In contrast to these findings however, a qualitative study by Reid et al., conducted in Canada, revealed a tendency to have unrealistic expectations of life expectancy and a lack of awareness and understanding regarding future health risks (17, 159).

2.14.2.1 Health behaviours

The engagement of adolescent CHD patients in common risk behaviours of adolescence seems low relative to their healthy peers. They are less likely to smoke, drink alcohol or use illicit drugs and are also less likely to be sexually active (137, 160). It should be noted however, that, while adolescents and young adults with CHD are less sexually active than their healthy peers (particularly true for adolescents), Reid et al. found that among those who are sexually active, there is engagement in potentially risky sexual behaviour (36% of young adults and 72% of adolescents), such as questionable birth control, using drugs or alcohol before sex, or two or more partners in the past 3 months. The authors therefore recommended sexual health be discussed with adolescents and young adults with CHD (160).

Overall however, the relatively low levels of engagement in risky behaviours by adolescents with CHD are encouraging given the negative health implications (17). However, it should be noted that these positive health behaviour choices are not always easy. Accepting their physical limitations (due to decreased stamina caused by CHD) and not being able to consume alcohol as freely as peers (due to medication side effects) can be challenging and isolating (161) and can even result in discrimination and bullying which in turn results in feelings of anger and exclusion (154).

As Sable et al. reported, qualitative and quantitative data on the quality of life of teenagers with CHD suggests that they face unique challenges on top of those typically faced during adolescence. By researching and attempting to understand the reasons behind adolescent perceptions of their health status and quality of life, it is hoped that appropriate education and support can be provided to them during their transition process (17).

2.14.2.2 Developmental Issues, Psychological adjustment and Quality of Life

As stated above, psychopathological development in children with CHD can emerge in the form behavioural disorders, both externalising and internalising (157). In a minority of cases, persistent cognitive and motor impairments can occur, such that long-term neurodevelopmental evaluations and interventions are necessary to provide early educational and therapeutic support (162). Research has been unable to agree upon definitive risk factors or determinants of developmental problems but researchers acknowledge genetic factors may contribute suggesting the need for early recognition of those potentially at risk. They also recommended further research to determine protective and risk factors linked to personality, environmental and relational aspects (163).

Following a systematic review and meta-analysis of the literature in relation to the psychological and cognitive functioning of children and adolescents with CHD, Karsdorp et al. found only older children and adolescents display increased risk of overall, internalizing and to a lesser extent externalising behavioural problems. More severe CHD was also related to lower cognitive functioning (specifically performance intelligence) and this decreased functioning remained fairly stable across different age groups (16).

Latal et al.'s systematic review was again focused on children and adolescents, in this case after open heart surgery. They commented on the lack of consensus regarding long-term psychological adjustment and health-related quality of life (164). While patients tend to self-report good outcomes, according to their parents a significant proportion experience psychological maladjustment. Parental reports of maladjustment were related to CHD severity and developmental delay (164). Studies also suggest impaired quality of life for some children, especially those with more severe CHD (164). Adolescents with CHD have shown poorer outcomes in relation to full-scale IQ, perceptual reasoning, working memory, visual perception, visuomotor integration, executive functions and all motor domains except static balance (162). On a more positive note, psychological adjustment was affected only in the peer relationship domain and quality of life was similar to healthy peers (162).

Portuguese studies have found a lifetime prevalence of approximately 20% for psychopathology in adolescent and young adult CHD patients with higher rates in female patients (165, 166). Female CHD patients and those with poor social support, poor academic performance or more complex CHD all do worse in relation to both psychosocial adjustment and quality of life (165). Specifically, females showed more somatic complaints, anxiety/depression, aggressive behaviour, attention problems, thought problems, internalisation and externalisation. Poor social support was associated with more withdrawal and social problems and poor school performance with anxiety/depression and attention problems (166). Gierat-Haponiuk et al. also found diminished quality of life and more frequent depression in young adults who had undergone surgical correction of a defect compared to healthy peers (104).

Overall, psychological adjustment and self-reported health related quality of life are similar in adolescents with CHD to those of adolescents in the general population (162). The percentage of adolescents with CHD who have emotional and behavioural problems is only slightly greater than in the general population (167). There is some evidence that females and those with more severe CHD and poorer social support or academic performance are more prone to maladjustment or lower quality of life (165, 166). To minimise psychological and behavioural difficulties, Karsdorp et al. have suggested that older children and adolescents with CHD may benefit from psychological interventions to reduce anxiety symptoms and depression (16).

2.14.2.3 Adolescents with CHD in Ireland and Northern Ireland

While there is no published research on living with CHD in Ireland, patients, parents and healthcare professionals have spoken to the media about some of the issues they encounter. For instance, a clinical nurse specialist has discussed the difficulty among young male patients in accepting advice to avoid heavy weightlifting and use of supplements like Creatine (74). Adolescents who were interviewed also spoke about struggling with giving up competitive sport ('devastating and aggravating'), exclusion from sport in general and PE (annoyance and frustration) and a sense of isolation (an adolescent from Northern Ireland referring to friendships built through sport). They did not report worry with regard to their future health, whereas one mother referred to herself as 'still traumatised' (74).

In addition to exercise, the clinical nurse specialist interviewed also described spending a lot of time on other lifestyle advice including diet, alcohol, recreational drugs, smoking and contraception (74). It is known that congenital heart disease increases risk of endocarditis, which is caused by bacterial infection. Those with CHD and the professionals responsible therefore need to be aware and take care to avoid infection during dental procedures (168) or if they decide to get any piercings or tattoos (169). All three adolescents interviewed in the article said that they prefer to keep their heart condition quiet and prefer not to talk about it. One reported really enjoying a teenage support group – the Children’s Heartbeat Trust (NI) - where they can socialise with others who understand their issues rather than discuss their heart conditions (74).

In the Republic of Ireland, Heart Children Ireland had a preliminary meeting of young adults with CHD. Margaret Rogers, the chief executive, told the *Irish Times* the main issues reported were ongoing healthcare, peer support and the determination to thrive, rather than just survive (74) and Heart Children Ireland had plans to launch a support group in Dublin for young adults aged 18-plus with CHD in last February. Again, a scientific study would be a valuable addition to knowledge in this area.

2.14.3 Young Adults

Berghammer et al. looked at the experiences of young adults (22-39 years old) as distinct from adolescents. These patients discussed their need to strike a balance between being different and not being different and revealing versus hiding their congenital heart disease (19). There seems to be a tendency to waver between wanting people to know about their condition and wanting to be ‘normal’ and like everybody else, and not talk about the disease (19). Patients feel this is related to the invisible nature of their condition. Feeling different often leads to feeling excluded socially and their strong wish to be healthy can lead young adult CHD patients to hide symptoms from healthcare personnel and even from themselves. Based on these findings, Berghammer et al. recommend applying a biopsychosocial model in the care of CHD patients (19).

A qualitative study exploring the experiences of young adults with congenital heart disease concluded that they require a structured, gradual and natural transfer process when moving to adult care (10). Healthcare needs to be organised so that care staff are able to provide information, preparation, education, continuity and accessibility. Also required is the continued support of parents. Finally, the young adults themselves must possess knowledge of their condition, be sufficiently mature and have the ability to take over responsibility (10).

2.14.4 Adult patients

As discussed above, feeling different is a feature of CHD in childhood, adolescence and young adulthood (18, 19). “Feeling different” has been identified as a central theme for adult patients living with congenital heart disease (20). As in children, it is the physical limitations and visible signs caused by heart defects which lead to this feeling and adult patients struggle both with themselves and their environment to be accepted as normal. Feeling different is influenced by the attitudes of the environment, health care and the patient’s personality and furthermore it is this feeling that determines the perceived impact of the disease on the patient’s daily life (20). Given this, achieving a sense of ‘normalcy’ has been identified as the most important process when dealing with patients suffering from CHD (20).

Even in adulthood, patients report doubt as to whether they should tell others about their heart disease (19). Adult patients also report shock and disappointment when they experience deterioration in their health after years of stability, a reminder that their CHD is a lifelong condition. A contradiction exists in that patients are in one way annoyed they were not forewarned but also did not want to feel different at the time or be told to be careful or expect decline. Perhaps it was this

denial and/or minimization that allowed them to enjoy a positive adolescence (18). Thus denial appears to be both adaptive and maladaptive for CHD patients with a shifting balance occurring throughout the lifecycle. It is currently unknown how patients in Ireland feel about this – would they like to know more or are they ‘just trying to get on with things and not let it take over their lives’?

Adult CHD patients have been shown to have healthier lifestyles than the general population (91). Adults with CHD smoked less, had more sports participation and less obesity (91). More negative outcomes were found in relation to education, employment and marital status however and the authors noted that this social disadvantage was present in all severity groups and primarily in young men (91). The ‘social burden’ reported by this study is challenged by the findings of a systematic review on quality of life in adult CHD patients (170). After reviewing 31 studies, the authors concluded that while quality of life was compromised in the physical domain compared to healthy peers, no differences were found in relation to the psychosocial and environmental/occupational domains.

Kovacs et al. reviewed the literature and found conflicting findings in relation to the emotional adjustment of adults with CHD. While most European studies suggested favourable outcomes in terms of emotional functioning, American studies seemed to indicate poorer emotional functioning (84). By examining the literature more closely however, they were able to conclude that interview data suggests more psychological disturbance than self-report data. This pattern has also been seen in adult survivors of childhood cancer (171) and may be due to denial and high achievement motivation leading to artificially inflated ratings of emotional state on self-report measures (90). It is also unclear what role sociocultural differences and disease severity may play in the emotional adjustment of adult CHD patients (84). On balance however, as is the case in adolescents with CHD, the prevalence of emotional and behavioural problems is only slightly greater in adult patients in comparison to the general population (167). This is not to underplay the importance of psychosocial adjustment - while problems may not be prevalent, where present they can have significant impact. As Dulfer et al. found in their study, patients with congenital heart disease rarely have depression, however, even minor depressive symptoms have been shown to have a stronger impact on quality of life than limitations of exercise capacity (119).

Future research

There is virtually no Irish research on inherited cardiac conditions or congenital heart disease. The international literature addressing the experiences of individuals and families living with these conditions may not be generalizable to Ireland in many cases. For instance, vastly different healthcare or insurance systems may mean some literature is not so applicable here and the same conclusions cannot be readily applied. Therefore, research to understand the Irish population and Irish context is required.

Primary research involving those living with inherited cardiac conditions and their families in Ireland is therefore needed and will be invaluable in assisting the Irish Heart Foundation and the Irish Health services in planning how they can best assist these individuals and their families now and in the future. Any such research should cover a range of ages, conditions and situations to ensure thorough coverage of all relevant facts of living with an inherited cardiac condition in contemporary Ireland. Both quantitative and qualitative methodologies should be considered, although qualitative methods are recommended initially for identifying the most important themes common to these patients. Quantitative studies could follow once these areas have been found. Screening or genetic testing and the stress it may cause has emerged as a big issue in the inherited cardiac conditions literature but in Ireland to date this has only been assessed cross-sectionally. In relation to young people living with heart defects, transition appears to be a crucial process. Transition within the

healthcare system, but also on a more personal level, with a shifting of responsibility from parent to child, appears to be a vital but often complicated process. Future research in each of these areas is required.

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