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Exploring How the Risk of Sudden Cardiac Death is Discussed in Families with a Diagnosis of a SADS Condition

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by

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Abstract

Sudden arrhythmic death syndrome (SADS), where death is secondary to cardiac arrhythmia, is associated with several cardiac ion channelopathies, including long QT syndrome and Brugada syndrome, as well as cardiomyopathies such as hypertrophic cardiomyopathy and dilated cardiomyopathy. Many of these conditions often present in childhood or adolescence. This study investigates how diagnoses of cardiac diseases associated with SADS are communicated within families. A questionnaire was distributed through cardiac disease-focused support groups and organizations. Data from 114 parents who have a child with a SADS condition were used for analysis. Based on the responses, parents explained the risk of SADS in a straightforward manner and related the risk to the importance of compliance with the prescribed treatment. Participants also found it difficult to determine and enforce lifestyle modifications, manage the families' emotional reactions, convey the seriousness of the information without scaring their children, and discuss the risk of SADS during these conversations. Concerns regarding disease progression, length and quality of life, and treatment failures and complications were also expressed. Healthcare providers, the Internet, other affected people, visual aids, and personal experience were all reported to be helpful for discussing the SADS condition with their children. Services and resources that were requested were children's support groups, a counselor or psychologist, and

child-oriented materials. Increased understanding of how families discuss children's diagnosis of SADS conditions will equip healthcare providers with the information to address parental concerns and help facilitate discussion of the condition between parents and their children.

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List of Abbreviations

AED ..... Automated External Defibrillator

ARVC ..... Arrhythmogenic Right Ventricular Cardiomyopathy

BrS .....Brugada Syndrome

CPVT .....Catecholaminergic Polymorphic Ventricular Tachycardia

DCM .....Dilated Cardiomyopathy

EKG .....Electrocardiogram

GINA..... Genetic Information Nondiscrimination Act

HCM ..... Hypertrophic Cardiomyopathy

ICD.....Implantable Cardioverter-Defibrillator

LQTS..... Long QT Syndrome

PCCD .....Progressive Cardiac Conduction Defect

SADS .....Sudden Arrhythmic Death Syndrome

SCD.....Sudden Cardiac Death

Chapter 1: Background

1.1 Overview of Cardiac Conditions

Sudden cardiac death (SCD) accounts for 50-100 deaths per 100,000 in North America and Europe every year (McGorrian et al., 2013). Sudden arrhythmic death is a specific type of SCD where sudden death is secondary to a cardiac arrhythmia. In cases of SCD where a structural heart defect cannot be found, the cause of death in these individuals is attributed to sudden arrhythmic death syndrome (SADS) (Vyas & Lambiase, 2013). Several studies have investigated the incidence of SADS; it has been reported to account for 0.16-0.24 deaths for every 100,000 people per year. However, the prevalence of SADS was found to be several times higher in younger populations; it is estimated to be 0.76 per 100,000 people per year in individuals aged 14-35 (McGorrian et al., 2013; Vyas & Lambiase, 2013).

A genetic predisposition can cause an increased risk for SADS in certain family members. Inherited cardiac disease has been found in up to 50% of families with a history of SADS (Vyas & Lambiase, 2013). Several diseases affect the electrophysiology of the heart without altering the actual cardiac structure. These conditions are collectively known as ion channelopathies and are associated with a risk of SADS. The most common are long QT syndrome (LQTS), Brugada syndrome (BrS), and catecholaminergic polymorphic ventricular tachycardia (CPVT), but others such as progressive cardiac conduction defect (PCCD), early repolarization syndrome, and short QT syndrome are also associated with a risk of SADS (Behr, 2010; Vyas & Lambiase,

2013). It is thought that these conditions are responsible for about 40% of all SADS cases (Behr, 2010). In about 10-20% of SADS cases, certain types of structural heart disease are found to be responsible. Hypertrophic cardiomyopathy (HCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), and dilated cardiomyopathy (DCM) are the some of the most frequent structural heart diseases that cause SADS (Behr, 2010).

Long QT syndrome (LQTS) is the most common ion channelopathy with a reported incidence ranging from 1 in 2,000 to 2,500 (Ackerman et al., 2011; Vyas & Lambiase 2013). It is characterized by a prolonged ventricular repolarization and a predisposition for a specific type of arrhythmia known as torsades de pointes (TdP), which leads to syncope, seizures, and SCD; this arrhythmia can begin as early as infancy. However, not everyone who has LQTS will become symptomatic in their lifetime. While it is not possible to predict a patient's prognosis, the probability of developing cardiac symptoms is partially dependent on a person's age, sex, and the length of the QTc interval. Within the symptomatic and untreated population, the mortality rate is about 50% (Ackerman et al., 2011).

Congenital LQTS is also marked by locus and allelic heterogeneity. Loss-of-function mutations in *KCNQ1* and *KCNH2*, which code for subunits of a cardiac potassium channel, are responsible for LTQ1 and LTQ2, respectively. LTQ3 is caused by gain of function mutations in *SCN5A*, which codes for a subunit in a cardiac sodium channel. Mutations in these three genes account for 70-75% of LQTS cases and are inherited in an autosomal dominant manner with a 5-10% *de novo* rate. (Ackerman et al., 2011; Bastiaenen & Behr, 2011). The other 25% of cases are due to a variety of mutations in nine other known genes as well as an unknown number of unidentified

genes. In addition, the other 25% of LQTS may be polygenic, and can display both autosomal dominant and autosomal recessive inheritance (Bastiaenen & Behr, 2011).

Management of LQTS mainly consists of pharmacotherapy with beta-blockers, life-style modifications, such as exercise restrictions, and avoidance of QT-prolonging drugs to reduce the risk of arrhythmia and syncope. High-risk patients may also have an ICD or pacemaker implanted (Ackerman et al., 2011). As genotype-phenotype correlations have emerged, medical management for LTQ1-3 has become partially dependent on the specific mutation discovered in each patient. Medical management with beta-blockers works best in patients with LQT1 and LTQ2; it is considered significantly less effective in LQT3 patients. The triggers that are most likely to cause patients to become symptomatic are also gene-specific. Exercise is a known risk factor for LTQ1; therefore, it is advised that these patients avoid competitive sports. Symptoms for patients with LQT2 can be caused by intense emotions and startling noises. In contrast, most patients with LQT3 become symptomatic while resting or sleeping (Napolitano, Bloise, Monteforte, & Priori, 2012). While these genotype-phenotype correlations have helped with symptom management, patients may experience symptoms more commonly associated with a different subtype of LQTS. Therefore, it is important to discuss all risk factors and possible symptoms with patients diagnosed with LQTS.

Brugada syndrome (BrS) is characterized by a conduction delay in the right ventricle as well as an elevated ST segment in the right precordial leads on an electrocardiogram (EKG) (Bastiaenen & Behr, 2011). The incidence is thought to be 1 in 5,000-10,000 in Western countries, but it is significantly more common in individuals with Asian ancestry (Ackerman et al., 2011). BrS also occurs about eight times more

frequently in men than in women (Wilde et al., 2002). The annual risk for SCD in BrS is lower than in LQTS and is thought to be between 1-2% per year (Napolitano et al., 2012).

Like LQTS, BrS displays reduced penetrance and variable expressivity. Some patients may never experience symptoms, while others may experience syncope, palpitations, and/or SCD. Symptoms generally present when the patient is resting or sleeping; however, patients usually do not become symptomatic until adulthood (McGorrian et al., 2013). BrS also displays locus and allelic heterogeneity; it is associated with over 250 mutations in 10 different genes. The most common genetic cause is a loss-of-function mutation in *SCN5A*, which is found in about 20% of cases. Like the majority of LTQS cases, BrS is inherited in an autosomal dominant manner (Bastiaenen & Behr, 2011; Napolitano et al., 2012). ICD implantation is the standard treatment for high-risk patients. Quinidine, an anti-arrhythmic drug, is currently being considered to treat medium or low-risk patients. Because fever can be a trigger for cardiac events, treating febrile illness with ibuprofen is another aspect of medical management. Avoiding medications that can exacerbate the ST elevation is also suggested for patients with BrS (Bastiaenen & Behr, 2011).

Catecholaminergic polymorphic ventricular tachycardia (CPVT) is characterized by an exercised or acute emotional-induced ventricular tachycardia, which in turn can cause syncope, cardiac arrest, and SCD (Ackerman et al., 2011; Napolitano et al., 2012). The exact incidence of this condition is unknown. Morbidity and mortality are relatively high for untreated patients with CPVT; they have a 79% risk of having a cardiac event by age 40 as well as a 30% chance of SCD as the first presenting symptom. The average age of symptom onset is eight years of age. (Ackerman et al., 2011; Napolitano et al., 2012).

Two genes are currently known to be associated with CPVT. They code for the ryanodine receptor protein RyR2 and the cardiac calsequestrin protein CASQ2. Both of these proteins are involved in the release of calcium from the sarcoplasmic reticulum (Napolitano et al., 2012). Mutations in *RYR2* are responsible for the autosomal dominant form of CPVT, which accounts for about 60% of cases. *CASQ2* mutations are less common (3-5% of cases) and cause the autosomal recessive form of CPVT (Ackerman et al., 2011). The primary treatment modality for CPVT patients is pharmacotherapy with beta-blockers. However, about 30% of patients still have arrhythmic events while taking these medications; ICD implantation has been suggested for these individuals (Napolitano et al., 2012).

Hypertrophic cardiomyopathy is the most common genetic heart disease; it affects 1 in 500 people and is characterized by variable cardiac hypertrophy, muscle fibrosis, and myocyte disarray within the cardiac muscle. Patients may also experience syncope, dyspnea, and cardiac arrest. Like many other cardiac conditions, HCM displays reduced penetrance and variable expressivity. Patients with a known genetic cause generally present with a more severe phenotype than patients with negative genetic testing results (Ackerman et al., 2011; Bos et al., 2014).

HCM is also known as a genetically heterogeneous condition. At least nine genes that code for cardiac myofilaments (sarcomeres) have been implicated in HCM pathogenesis, and most of the mutations found to date are unique to each family and inherited in an autosomal dominant manner. The most commonly found mutations are in *MYBPC3* and *MYH7*; they each account for 25-33% of all cases (Ackerman et al., 2011). Several studies have attempted to make genotype-phenotype correlations with the several

sarcomere genes associated with HCM. While no mutation-specific correlations have been made, patients carrying a sarcomere mutation tended to present with a more severe phenotype; they generally presented at an earlier age, have a greater degree of hypertrophy, and have a greater frequency of SCD and family history of HCM (Lopes, Rahman, & Elliott, 2013). The medical management plan for patients with HCM consists of symptom treatment, surveillance and prevention of complications, and may include pharmacotherapy, surgery, and pacemaker/ICD implantation (Cirino & Ho, 2008). In a subset of patients, HCM is a feature of another genetic condition. There are several muscular dystrophies as well as metabolic and mitochondrial disorders associated with cardiomyopathies like HCM or DCM. Although many patients with these disorders are at a high risk for arrhythmia and may require a pacemaker or ICD, treatment would be largely dependent on the specific diagnosis (Gilbert-Barness, 2004)

Arrhythmogenic right ventricular cardiomyopathy is a type of heart disease with a reported incidence of 1 in 1,000-1,250 people that results in the breakdown of the myocardium in the right ventricle. It is characterized by ventricular arrhythmia, syncope, and an increased risk for heart failure and/or SCD; however, due to reduced penetrance and variable expressivity, only about half of mutation carriers become symptomatic (McNally, MacLeod, & Dellefave-Castillo, 2005). This is an important factor to consider when counseling patients about their risk of arrhythmia and SCD. Exercise can be a possible trigger for patients with AVRC (Janzen et al., 2014). Most forms of AVRC are autosomal dominant; however, digenic and autosomal recessive forms do exist. Regardless of the inheritance pattern, AVRC displays both clinical and allelic heterogeneity. The majority of genes known to be responsible for this condition are

desmosomal proteins, which are involved in cell-cell adhesion. Mutations in these genes account for 30-70% of AVRC cases (Ackerman et al., 2011). Treatment for ARVC is similar to therapy for HCM. It is focused on minimizing syncope, cardiac arrest, and SCD with pharmacotherapy and ICD implantation (McNally et al., 2005).

Dilated cardiomyopathy (DCM) is characterized by enlargement of the left ventricle and systolic dysfunction, or reduced contractility. Common symptoms include arrhythmias, heart failure with congestion, fatigue, and dyspnea. This condition is also characterized by age-dependent penetrance with the age of onset varying from infancy to adulthood. DCM is thought to be more common than HCM; however, the exact incidence is unknown (Hershberger & Morales, 2007). Most genetic forms of DCM are inherited in an autosomal dominant fashion, but autosomal recessive and X-linked inheritance patterns have also been seen. Over 30 genes have been implicated in DCM; however, each gene accounts for less that 5% of the DCM cases (Ackerman et al., 2011). Treatment is similar to the previously described cardiomyopathies. The main modalities of treatment include pharmacotherapy with antiarrhythmic drugs, ICD or pacemaker implantation, and heart transplantation for advanced heart failure.

1.2 Diagnosing Cardiac Conditions

While some affected individuals will have obvious and distinct clinical features that lead to a clear diagnosis, most cases are not this straightforward. Since these ion channelopathies and cardiomyopathies show reduced penetrance and variable expression, not all mutation carriers will present with a phenotype indicative of a cardiac condition. It often requires a "perfect storm;" a combination of genetic predisposition and environmental factors, including a trigger, to set off a severe arrhythmic event that would

be suggestive of a cardiac disease (Janzen et al., 2014). Therefore, a variety of tests and evaluations are used to help detect affect individuals with more subtle phenotypes.

A thorough medical history can help narrow the differential diagnosis when screening for cardiac disease. Important elements of the medical history include detailed information about any previous episodes of syncope or palpitations, such as what the patient was doing when (s)he become symptomatic as well as any medication (s)he was taking. A family history of syncope, cardiac arrhythmia, seizures, dizziness, sudden cardiac arrest, or SCD can also be strongly indicative of an inherited cardiac disease.

A medical examination is another important part of the evaluation when screening for cardiac disease, and can include several tests. In addition to a resting electrocardiogram (EKG), signal-averaged EKGs, provocative stress EKGs, Holter monitors, and implantable loop monitors are also used to detect cardiac arrhythmias. Signal-averaged EKGs look at about 400 heartbeats and can be used to detect less obvious arrhythmias that may be missed on a resting EKG. A provocative stress test is similar to a resting EKG, but instead of remaining still, the electrical activity of the heart is recorded before, during, and after either exercise of varying intensity or administration of an antiarrhythmic drug. This test makes it possible to pick up arrhythmias that only present when physically stressed and is the primary diagnostic tool used for CPVT (Behr, 2010; Janzen et al., 2014). A holter monitor is a portable EKG that records the electrical activity of the heart for 24 to 48 hours, and can be used to detect arrhythmias that are intermittent. An implantable loop monitor is a small device that is placed under the skin and can be used to record a patient's heart rhythm for up to three years. These devices are useful if a patient's symptoms occur less frequently (Mofrad, 2012). An

echocardiogram, or an ultrasound of the heart, is another method used to identify cardiac conditions. It is useful when determining if there is any structural damage, and is the gold standard for diagnosing HCM, DCM, and ARVC (Behr, 2010). A cardiac MRI is a second diagnostic tool that allows physicians to visualize the structure of the heart for diagnosing certain cardiac conditions (Janzen et al., 2014).

Genetic testing is an additional technique that is utilized to help diagnose cardiac disease. In families where a member has suffered from SCD, a molecular autopsy can be performed. If a familial mutation has not been previously identified in a family, DNA testing for a number of genetic mutations that cause inherited cardiac conditions can be done on DNA of the deceased individual. If a pathogenic mutation is found, cascade screening of at-risk relatives can help identify other mutations carriers in the family. Conversely, if a familial mutation has already been identified, genetic testing for that specific change in the DNA can be done to diagnose the cause of death (Behr, 2010).

1.3 Implications of Genetic Testing

Genetic testing for ion channelopathies and cardiomyopathies is also performed on living patients when cardiac disease is suspected. Before genetic testing is completed, however, there are several logistical and psychosocial considerations that need to be addressed. One aspect to consider is the benefits and limitations of genetic testing. Confirming a clinical diagnosis with genetic testing can have prognostic and therapeutic implications that will aid in medical management, but genetic testing is not an infallible technique. While genetic testing detects mutations in the majority of patients for some conditions like LQTS, genetic testing for other conditions yields a much lower positive mutation rate. For example, mutations are only found in 20-30% of clinically diagnosed patients with Brugada syndrome (Bastiaenen & Behr, 2011). Therefore, a

negative genetic test result does not necessarily rule out a genetic cause for a patient's clinical features. A second issue that must be discussed is the possibility of genetic discrimination. Although the Genetic Information Nondiscrimination Act (GINA) was passed in 2008 to prevent genetic discrimination in the workforce and health insurance, there are other sectors in which genetic discrimination is not prohibited. These areas include life insurance, long-term care insurance, and disability insurance. Therefore, patients should be counseled to consider obtaining these types of insurance before being tested.

If a patient's genetic testing results are positive for a cardiac disease-causing mutation, family cascade screening is strongly recommended (Ackerman et al., 2011). Mutation-specific screening for at-risk family members is suggested for several ion channelopathies and cardiomyopathies, including LQTS, BrS, CPVT, HCM, ARVC, and DCM as well as several other conditions (Ackerman et al., 2011). Family screening can help relieve uncertainties regarding an individuals' carrier status as well as guide medical management. An asymptomatic family member who is a mutation carrier can start taking preventive measures to reduce their risk of syncope, arrhythmia, and SCD. However, since most of these conditions exhibit reduced penetrance and variable expressivity, some mutation carriers will remain asymptomatic throughout their life (Ackerman et al., 2011). Therefore, each individual must consider the implications of both undergoing testing and remaining unaware of their carrier status before making a decision. People who choose to undergo genetic testing list several motivations for their decision, including to find an explanation for family history of sudden death, to confirm a clinical diagnosis, to aid in medical management decisions, to alleviate concerns about risk to other family members,

and to comply with physicians' or relatives' recommendations. A lack of information, denial, and fear were reasons people had reported in their decisions not to pursue genetic testing (Erskine et al., 2014).

Many individuals, regardless of their age, experience psychosocial consequences of discovering they are a mutation carrier for a cardiac disease. One study found that patients experienced significantly increased levels of heart-focused anxiety after learning about their genetic diagnosis of LQTS or HCM. The three aspects of anxiety that they focused on are avoidance of activities thought to trigger cardiac symptoms, increased attention toward cardiac activity, and fear regarding heart sensations (Hamang et al., 2012). Even though these feelings seemed to persist over an extended period of time, their study showed that patients who underwent genetic counseling experienced reduced levels of cardiac avoidance and attention. These findings indicate that providing accurate information about their condition as well as psychosocial support can help lower their heart-focused anxiety.

Numerous individuals with inherited cardiac disease present with symptoms during childhood; therefore, a significant portion of the population undergoing genetic screening is under the age of 18. In addition to the topics that should be addressed for all patients, there are special considerations that should be taken into account when testing children. Several case studies have been done to investigate the challenges health professionals may encounter while counseling children and adolescents regarding genetic testing (Callard, Williams, & Skirton, 2012; Cohen, Stolerman, Walsh, Wasserman, & Dolan, 2012). Medical professionals must balance the wishes of the parents, child, and the medical necessity when discussing genetic testing.

Adolescents' cognitive ability and maturity level are two factors that may make counseling them more difficult. When children refuse to be tested, it can be difficult to determine if it is because they are misinformed, are unable to consider the long-term implications of their decision, or are choosing to live with the risk of potentially being a mutation carrier. They may also be unwilling to comply with the lifestyle changes that are recommended for mutation carriers (Cohen et al., 2012). When the parents and/or the child are opposed to genetic testing, it is important to ensure that they understand the diagnostic, prognostic, and therapeutic benefit that genetic testing may provide.

Parents may also be resistant to testing their children. According to Cohen, Stolerman et al., many parents who do not wish to have their children tested are concerned about the psychological impact testing would have on their children and that it would outweigh the benefits of knowing their genetic status (2012). Although these concerns are understandable, Meulenkamp et al., have shown that learning about their carrier status does not affect children as significantly as some parents think it will (2008). They found that most children became well-adjusted to their diagnosis and were knowledgeable about the genetic nature and lack of cure for their condition. Participants who were having trouble coping with their condition seemed less informed about their diagnosis and the steps that should be taken to reduce their risk of SADS.

The authors also reported implications for counseling based on their data. First, they emphasized the necessity of children understanding their diagnosis as well as the implications of their carrier status. Second, they recommended that children should have a realistic understanding regarding the how controllable the condition is. The authors found that providing information on what steps should be taken to reduce their risk and

why these measures work will help reduce worries in carrier children. The third implication is the necessity of parental support as they makes choices about how to best protect their children without imposing excessive limitations on their activities.

1.4 Communicating Genetic Risk to Children

Previous research has made it clear that children are not adults in smaller form; they are still undergoing cognitive development and therefore require information to "be tailored to their social, emotional, and cognitive development"(Sullivan & McConkie-Rosell, 2010, p.231). For children to understand complex concepts such as genetic conditions, the information must be simplified to the appropriate level and communicated using appropriate emotional tones in an open style of communication (Sullivan & McConkie-Rosell, 2010). Although minimal research has been published on how parents communicate risk information about cardiac diseases associated with SADS (Mangset & Hofmann, 2014), studies have been done to investigate how parents communicate diagnoses and related risk information to their children.

These studies have similar findings regarding the communication pattern between parents and their children as well as the potential barriers that may affect communication. One study, completed by McConkie-Rosell and her colleagues, investigated how parents disclose risk information to their daughters who are potential carriers for Fragile X syndrome, a genetic condition that causes developmental disabilities primarily in males (2011). When asked, the girls stated that they desired a resilient communication style. They preferred "having an 'actual conversation,' the information to be staged, given with reassurance, normalized, and that parents be truthful, honest and knowledgeable about the genetic information" (McConkie-Rosell, Del Giorno, & Heise, 2011, p.59). This finding correlates with the conclusions made from another study of families with children

diagnosed with LQTS; the majority of parents felt that it was important to know their children's carrier status and be open and honest with the information (Mangset & Hofmann, 2014). Another study, which explored how genetic risk information was discussed for a variety of Mendelian disorders, found that when information about their condition was disclosed gradually from an early age, children were more able to cope with the condition (Metcalfe, Plumridge, Coad, Shanks, & Gill, 2011). Not only did they have a better understanding of their condition in the long run, but they were also able to learn about the implications with less of a shock. These findings emphasize the importance of disclosing information about a child's diagnosis in an appropriate way.

Even though many parents and children express desire for early and direct communication, it does not always occur. One fourth of the girls in the study of families with Fragile X reported that they became aware of their personal risk for carrying a mutation exclusively through indirect communication (McConkie-Rosell, Heise, & Spiridigliozzi, 2009). Metcalfe and her colleagues also found that most parents delayed discussing their genetic risk information with their children for as long as possible (2011). After interviewing the parents, the authors of these studies proposed several potential barriers to the open communication the families preferred. The need to protect their children, the shock of the diagnosis, and feelings of guilt, fear, and grief were all reasons parents delayed or avoided discussing genetic risk information with their children (Metcalfe et al., 2011). Other possible barriers to communication include hesitation about when to disclose, how to phrase the complex information and the implications for the child's future as well as uncertainty regarding what the child will be able to understand (McConkie-Rosell et al., 2011). Batte et al. (2015) made similar findings

when they surveyed families with a history of HCM. They identified an accurate understanding of the risk for other family members as a propeller of familial communication and dysfunctional family dynamics as a factor that may hamper communication within a family. Previous articles have recommended several activities that healthcare professional can partake in to help parents with the disclosure process. Discussing communication styles the family currently uses, brainstorming possible questions the children may ask, exploring how to describe the information using language the child will comprehend, and allowing the parents to practice dialoging the conversation are all helpful suggestions (McConkie-Rosell et al., 2011). However, it is not known what type of guidance is most wanted by parents of children who have cardiac conditions with a risk of SADS.

Some of the parents expressed a desire for more instruction on how to talk to their children about genetic risk information during the interview process. One of the fathers stated: "Perhaps the next step for us now is to get some guidance on how to talk about it without making her very scared" (Mangset & Hofmann, 2014). Surveying parents whose children have a variety of cardiac diseases associated with SADS will provide more detailed information on what exactly families who are adapting to these conditions need from healthcare professionals.

CHAPTER 2: Exploring How the Risk of Sudden Cardiac Death is Discussed in Families with a Diagnosis of a SADS Condition[1]

2.1 Abstract

Sudden arrhythmic death syndrome (SADS), where death is secondary to cardiac arrhythmia, is associated with several cardiac ion channelopathies, including long QT syndrome and Brugada syndrome, as well as cardiomyopathies such as hypertrophic cardiomyopathy and dilated cardiomyopathy. Many of these conditions often present in childhood or adolescence. This study investigates how diagnoses of cardiac diseases associated with SADS are communicated within families. A questionnaire was distributed through cardiac disease-focused support groups and organizations. Data from 114 parents who have a child with a SADS condition were used for analysis. Based on the responses, parents explained the risk of SADS in a straightforward manner and related the risk to the importance of compliance with the prescribed treatment. Participants also found it difficult to determine and enforce lifestyle modifications, manage the families' emotional reactions, convey the seriousness of the information without scaring their children, and discuss the risk of SADS during these conversations. Concerns regarding disease progression, length and quality of life, and treatment failures and complications were also expressed. Healthcare providers, the Internet, other

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affected people, visual aids, and personal experience were all reported to be helpful for discussing the SADS condition with their children. Services and resources that were requested were children's support groups, a counselor or psychologist, and child-oriented materials. Increased understanding of how families discuss children's diagnosis of SADS conditions will equip healthcare providers with the information to address parental concerns and help facilitate discussion of the condition between parents and their children.

2.2 Introduction

Sudden arrhythmic death is a subtype of sudden cardiac death (SCD) where death is secondary to a cardiac arrhythmia. An individual's death is attributed to sudden arrhythmic death syndrome (SADS) in cases of SCD where a structural heart defect is not present (Vyas & Lambiase, 2013). Numerous studies have investigated the incidence of SADS; the estimated prevalence of SADS ranges from 0.16-0.24 per 100,000 people per year in the general population to 0.76 per 100,000 people per year in individuals aged 14-35 (McGorrian et al., 2013, Vyas & Lambiase 2013).

A genetic predisposition for cardiac disease has been found in up to 50% of families with a history of SADS (Vyas & Lambiase, 2013). A subset of cardiac conditions, collectively known as ion channelopathies, affect the electrophysiology of the heart without altering the actual cardiac structure. Long QT syndrome (LQTS), Brugada syndrome, and catecholaminergic polymorphic ventricular tachycardia (CPVT) are three of the more common ion channelopathies associated with a risk of SADS (Behr, 2010; Vyas & Lambiase, 2013). As a group, ion channelopathies are thought to be responsible for approximately 40% of all SADS cases (Behr, 2010). Structural heart diseases, such

as hypertrophic cardiomyopathy (HCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), and dilated cardiomyopathy (DCM) have also been associated with SADS and are thought to be responsible for 10-20% of SADS cases (Behr, 2010).

While the symptoms patients experience vary among diseases, there is significant overlap in the above conditions. Both the ion channelopathies and the cardiomyopathies are characterized by arrhythmia, syncope, seizures, fatigue, and cardiac arrest (Ackerman et al., 2011). In addition, the cardiomyopathies are characterized by specific structural abnormalities and have a risk for congestive heart failure. Most mutations are inherited in an autosomal dominant manner, but other inheritance patterns do exist for these conditions. Both classes of cardiac diseases also display reduced penetrance and variable expressivity. Therefore, not everyone who carries a mutation will develop symptoms. The morbidity and mortality rate is dependent upon a multitude of factors. In addition to the clinical diagnosis and specific mutation, a patient's age, sex, and lifestyle choices can also influence their risk for developing symptoms (Ackerman et al., 2011). There is also a notable range in mortality rates for the above conditions. The annual risk of sudden cardiac death is thought to be 1-2% for Brugada syndrome (Napolitano et al., 2012). In contrast, the mortality rate for the untreated LQTS population may be as high as 50% (Ackerman et al., 2011).

There are similarities in the suggested medical management for the above cardiomyopathies and ion channelopathies as well. Treatment mainly consists of pharmacotherapy with beta-blockers and other medications, lifestyle modifications (such as exercise restrictions), and avoidance of drugs that increase the likelihood of arrhythmic events. High-risk patients may also have an implantable cardioverter defibrillator (ICD)

or a pacemaker implanted (Ackerman et al., 2011; Napolitano et al., 2012). Some patients with cardiomyopathy may require a heart transplant if they develop advanced heart failure (Ackerman et al., 2011).

Medical professionals currently utilize a variety of tests and evaluations to diagnose these conditions, including documentation of a detailed family and medical history; a physical exam, consisting of an electrocardiogram, echocardiogram, and cardiac MRIs; and genetic testing (Behr, 2010; Janzen et al., 2014).

Many individuals with inherited cardiac disease present with symptoms during childhood; therefore, parents often find themselves having to explain the diagnosed cardiac condition to their children. Previous research has illustrated that children are still undergoing cognitive development and therefore require information "be tailored to their social, emotional, and cognitive development" (Sullivan & McConkie-Rosell, 2010, p.231). In order for children to understand complex concepts such as genetic conditions, the information must be simplified to the appropriate level and communicated using appropriate emotional tones in an open style of communication (Sullivan & McConkie-Rosell, 2010). However, minimal research has been published on how parents communicate risk information about cardiac diseases associated with SADS (Mangset & Hofmann, 2014). Surveying parents of these children would provide insight into this information, which can then be used to equip healthcare providers in helping parents have these crucial conversations with their child. Our study was expected to fill a gap in the current knowledge that will benefit cardiologists, genetic counselors, and other healthcare professionals, as well as the patients and their parents, as the data from this study will

allow healthcare professionals to better understand how to address these issues with the parents.

The primary objectives of this study included investigating how parents communicate with children about the children's diagnoses of cardiac diseases associated with SADS, exploring how the risk of sudden cardiac arrest is discussed between the parents and their affected children, and determining which aspects of these conversations parents find most difficult to communicate to their child. We hypothesize that numerous factors, including the children's specific diagnosis as well as the families' experiences with the condition and its associated symptoms have influenced how parents communicate their children's diagnosis to them.

2.3 Materials and Methods

This research study collected quantitative and qualitative data from parents of children who have a diagnosis of a cardiomyopathy associated with SADS. Participants were recruited through various organizations and Facebook groups targeted to families with children affected by the previously described conditions. Individuals over the age of 18 who had at least one child with a cardiomyopathy associated with a risk of SADS and could comprehend the medical & technical information in the questionnaire were eligible to participate in this study.

An invitation to participate (Appendix C) as well as a link to the survey (Appendix D) hosted on surveymonkey.com was distributed through several avenues. The SADS Foundation, Hypertrophic Cardiomyopathy Association, and the Cardiomyopathy Foundation were contacted via email to explain the purpose of the study and inquire if they were willing to invite their members to participate (Appendix A). All

three organizations were willing to invite their members to participate and did so via their respective electronic mailing lists. In addition, a link to the invitation to participate letter and survey were posted on various Facebook groups (Appendix B). The links were posted in September and October of 2014, and were available until December, 2014.

The survey consisted of multiple choice and open-ended questions that inquired about demographic information as well as the diagnostic process for their children's cardiomyopathy, how both the diagnosis and the risk of SCA is discussed between the parents and their children, and what aspects of these conversations are most difficult. Responding to each question was voluntary, which allowed participants to skip questions they did not wish to answer. Upon completion of the survey, participants were invited to provide their contact information to enter their name in a drawing for a $25 gift card to a location of their choice.

Quantitative analysis was performed using statistical analysis software (SAS) base 9.4. Fisher's exact test with and without Monte Carlo estimates as well as the extended Cochran-Mantel-Haenszel correlation statistic were used to identify significant relationships within the survey group. A 0.05 level of significance was used for all analyses. In addition, frequencies and percentages were calculated for each question. Qualitative data was analyzed by the principal investigator to identify recurring themes using Grounded Theory methods. This research study was approved by the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC, in August of 2014.

2.4 Results

2.4.1 Participant Demographics

A total of 114 participants completed the survey, all of which met the eligibility criteria. Respondent demographics are displayed in Table 2.1 and 2.2. The majority of participants were Caucasian (*n* = 95, 83%) females (*n* = 98, 86%) between the ages of 30 and 49 (*n* = 78, 68%) who had completed at least some college (*n* = 88, 77%) and were married (*n* = 78, 68%).

Table 2.1 Participant Demographics

		Frequency (N = 114)	Percentage (%)
Gender			
	Female	98	86
	Male	6	5
	Prefer not to answer	10	9
Age			
	<20	0	0
	20-29	5	4
	30-39	33	29
	40-49	45	40
	50-59	17	15
	60-69	4	3
	>70	0	0
	Prefer not to answer	10	9
Education level			
	Less than high school degree	0	0
	High school degree or equivalent	16	14
	Some college, but no degree	22	19
	Associate degree	13	11
	Bachelor degree	28	25
	Graduate degree	25	22
	Prefer not to answer	10	9
Ethnicity			
	American Indian or Alaskan Native	2	2
	Asian or Pacific Islander	1	1
	Black or African American	0	0
	Hispanic or Latino	3	3
	White/ Caucasian	95	83
	Prefer not to answer	13	11
Relationship status			
	Married	78	68
	Widowed	1	1
	Divorced	5	4
	Separated	4	4
	Domestic partnership or civil union	4	4
	Single	10	9
	Prefer not to answer	12	10

The majority of participants' children had either HCM (*n* = 61, 54%) or LQTS (*n* = 39, 34%). The most common age range at diagnosis was zero to three years (*n* = 32, 28%), followed by 10 to 12 (*n* = 19, 17%) and 13 to 15 (*n* = 19, 17%). The majority of children were between 10 and 18 years of age at the time the survey was completed, with the most commonly selected age range being 13 to 15 years. There were a relatively equal number of reported male (*n* = 58, 51%) and female (*n* = 56, 49%) children. For participants who had multiple children affected with a cardiomyopathy, data is displayed for their first child diagnosed (Table 2.2).

Table 2.2 Children Demographics

		Frequency (*N* = 114)	Percentage (%)
Gender			
	Male	58	50
	Female	56	50
Diagnosis			
	LQTS	39	34
	Brugada	5	4
	ARVC	1	1
	HCM	61	54
	DCM	2	2
	CPVT	2	2
	Unknown	2	2
	Other	2	2
Age at diagnosis			
	0-3	32	28
	4-6	6	5
	7-9	16	14
	10-12	19	17
	13-15	19	17
	16-18	13	11
	19-21	4	4
	22+	5	4
Current age			
	0-3	8	7
	4-6	8	7
	7-9	9	8
	10-12	20	18
	13-15	24	21
	16-18	15	13
	19-21	11	10
	22+	13	11
	Deceased	6	5

2.4.2 Experience with Symptoms and Treatment

The most commonly selected first presenting symptom was arrhythmia (n = 23, 21%), followed by syncope (n = 11, 10%). All other symptoms were experienced at the presentation of the condition in fewer than 10% of cases. Notably, 43% of the participants reported that their first child (if multiple children are affected) is asymptomatic. When questioned about the symptoms experienced by their child(ren) to date, arrhythmia (n = 45, 40%), dizziness (n = 43, 38%), and syncope (n = 27, 24%) were the most commonly reported symptoms. Lifestyle modifications (n = 72, 63%) and medications (n = 71, 62%) were the most frequently selected treatment options. ICDs and pacemakers were utilized by 27% and 9% of the respondents' children, respectively (Table 2.3)

Table 2.3 Symptoms and Treatment

		Frequency	Percentage (%)
First presenting symptom		n = 109	
	Syncope	11	10
	Arrhythmia	23	21
	Sudden Death	6	6
	Seizure	1	1
	Heart murmur	9	8
	Shortness of breath	3	3
	Asymptomatic	47	43
	Other	9	8
Symptoms experienced to date		N = 114	
	Syncope	27	24
	Arrhythmia	45	40
	Seizure	11	10
	Sudden Death	6	5
	Sudden Cardiac Arrest	7	6
	Dizziness	43	38
	Shortness of Breath	6	5
	None	13	11
Current treatment		N = 114	
	Medication	71	62
	Lifestyle Modifications	72	63
	Pacemaker	10	9
	ICD	31	27
	Other	11	10

2.4.3 Parental Concern Regarding Their Children's Cardiomyopathy

A total of 98 participants responded when asked the question, "What is your biggest concern for your child(ren) regarding their condition?". The majority of participants reported concerns were related to disease progression, especially SCA. Concerns regarding their children's length and quality of life, specifically the number of medical appointments, sports restrictions, and the children's emotional well being, were also commonly expressed. A third set of concerns surrounded their children's treatment. Respondents were worried about the lack of compliance by their children, as well as faulty treatments or treatment complications.

2.4.4 Communication Regarding the Diagnosis

Participants were questioned about who was involved in explaining the diagnosis to the children, the time frame that was taken to explain the diagnosis, and what topics were focused on in initial and subsequent conversations. The mothers of the children were the most frequently involved in the initial explanation (n = 90, 79%), followed by the physicians (n = 75, 66%), and then the fathers (n = 51, 45%). A significant correlation was found between the diagnosis and whether or not the father was involved in the explanation to the child (p =.0225) through the use of Cochran-Mantel-Haenszel statistics. While close to 60% of fathers were involved in the explaining a diagnosis of LTQS to their children, less than a third of fathers who had children diagnosed with HCM were involved in the explanation.

The majority of parents explained the diagnosis over a period of months or years (n = 62; 54%); however, a notable number of parents reported that the initial explanation occurred in a matter of days (n = 41, 41%). Utilization of the Monte Carlo estimate for

Fisher's exact test revealed a statistically significant association between the age of diagnosis and the time frame of the initial explanation ($p < .0001$). Parents were more likely to stretch the explanation over months or years if the child was diagnosed at a younger age. Approximately 10% of respondents selected two or more time frames, so the total percentage for the time period of the diagnosis does not add up to 100 percent. A third of the participants stated that the children's diagnoses were not discussed regularly; the other respondents reported discussing it daily, weekly, and monthly in approximately equal numbers (Table 2.4)

Table 2.4 Explanation of the Diagnosis

		Frequency	Percentage (%)
People involved in the explanation of the diagnosis		*N = 114*	
	Mother	90	79
	Father	51	45
	Other Relative	8	7
	Physician	75	66
	Genetic Counselor	19	17
	Other	5	4
Time period of explanation of diagnosis		*n = 101*	
	Days	41	41
	Weeks	15	15
	Months	28	28
	Years	34	34
Frequency of discussion		*n = 102*	
	Daily	21	21
	Weekly	23	23
	Monthly	27	27
	Not discussed regularly	34	33

The majority of topics were discussed in similar frequencies in the initial and subsequent conversations. Possible symptoms and medical management were the most commonly discussed topics in both the initial and follow-up conversations (60%-72%). The most notable difference between the initial and follow-up conversations was the 36% increase in the number of participants that discussed the genetic aspect of the conditions

in subsequent conversations when compared with the initial explanation. Respondents who selected "other" for both the initial and follow-up conversations mainly discussed topics related to medical management, such as participation in sports and surgeries. Figure 2.1 displays the percentage of participants that discussed each of the topics during the initial explanations and following conversations.

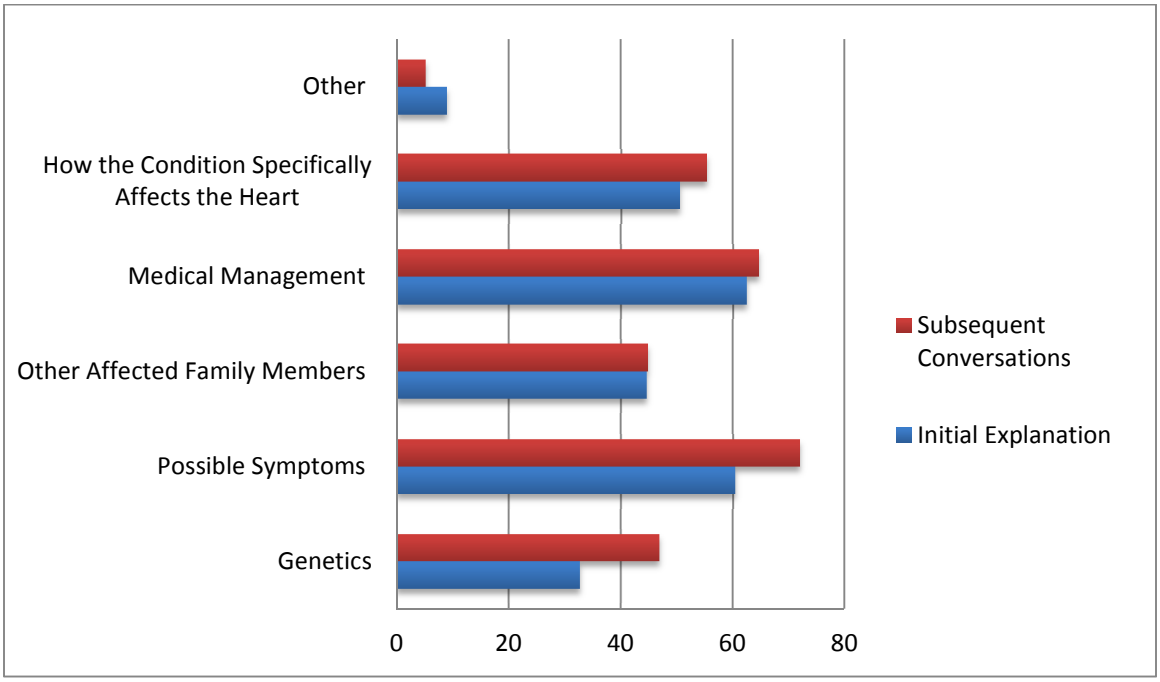

Figure 2.1 **Topics Discussed During Initial Explanation and Subsequent Conversations**

Statistical analysis was conducted to investigate if any significant relationships existed between what was discussed in the initial and follow-up conversations and the children's age at diagnosis, symptoms experienced to date, and the parents' understanding of their children's risk of SCA. The use of Cochran-Mantel-Haenszel Statistics revealed a statistically significant relationship between the child's age at diagnosis and whether or not genetics (p = .0331) and how the condition specifically

affects the heart (p = .0151) were discussed in initial conversations**.** As the age of diagnosis increased, parents were more likely to discuss the genetic aspect of the condition. While there was no linear relationship between the age of diagnosis and the frequency at which parents discussed how the condition specifically affects the heart, parents were most likely to describe this aspect of the condition when their child was diagnosed between the ages of seven and 15.

Utilization of Fisher's exact test revealed statistically significant associations between what was discussed in subsequent conversations and whether or not their children were symptomatic. Symptoms (p = .0053), how the condition affects the heart (p = .0192), medical management (p = .0195), and other family members with the same condition (p = .0296) were all more likely to be discussed in follow-up conversations if the participants' children were symptomatic.

Free-response questions were analyzed to identify what triggers these conversations and what aspects of these conversations were most difficult. The majority of participants reported triggers related to medical management, such as limitations from physical activity, an appointment with the cardiologist, medication, or an ICD implantation. Physical symptoms felt by their children, the psychosocial and emotional impact of the condition, and questions or comments made by the children were three other commonly reported triggers. Furthermore, a significant correlation (p = .0115) between the diagnosis and frequency of children's questions and comments being reported as a trigger was identified using Monte Carlo estimate for Fisher's exact test. While over 20% of parents whose children have LQTS reported it as a trigger, less than five percent of parents whose children have HCM specifically mentioned it as a trigger

for conversations regarding the diagnosis. No other associations between the reported triggers and the child's diagnosis were found.

Four main answers were identified from the parent's responses to the question, "What were the more difficult aspects of these conversations with your child?" Approximately one half of participants stated that discussing medical management, especially surgery and lifestyle stages, were most difficult for them. One mother reported:

> *Lifestyle modifications [have]been the hardest!! Both of my children love, love, love to play competitive sports. When the Dr. told both of my children they could not play it was the beginning of the emotional rollercoaster. My son whom loves football cried for hours. My daughter who loves volleyball was simply a wreck.*

A second set of answers was related to the psychosocial and emotional implications of the condition. In addition to their personal concerns, some participants stated that their children were experiencing anxiety attacks or struggling with "being different." One parent reported that her child was prescribed Zoloft to help manage her fear of her ICD firing. Multiple respondents also mentioned struggling with balancing information about the condition when talking with their children. In addition to having trouble explaining the condition in an age-appropriate way, parents found it difficult to "foster appropriate concern" without making their children constantly worry. Lastly, several parents said that they struggled with discussing the possible symptoms, specifically SCA. While there is not sufficient evidence that a statistically significant correlation between the diagnosis and reported difficult aspects of these conversations exists, the comparison between the diagnosis and whether or not the parent experienced

difficulty discussing medical management trends toward significance (p = .0586) and may warrant further investigation.

2.4.5 Risk of SADS

A total of 112 participants responded to the questions regarding their understanding and level of concern regarding their children's risk of SADS. Most respondents stated that they thought their children either had a low (n = 42; 38%) or moderate (n = 46; 41%) risk of SADS. However, the majority of parents (n = 59; 53%) expressed that they were very concerned about their children's risk of sudden death.

Four participants stated that they were not concerned about their children's risk of SADS. Of these respondents, two of them stated that they understood their children as being not at risk for SADS. Both of the children of these parents had a diagnosis of HCM, were diagnosed in between the ages of 13 and 18, are currently 16 or older, have only experienced either arrhythmia or a heart murmur, and are currently being treated with either medication or an ICD. The other two participants who expressed no concern for their children's risk of SADS reported their understanding of the risk for SADS to be either low or moderate risk. For the parent who selected low risk, her children have a diagnosis of Brugada, are asymptomatic, and are not being treated for their condition. For the parent who selected moderate risk and no concern, his or her children have a diagnosis of LQTS; have experienced syncope, arrhythmia, seizures, and dizziness; and are being treated with medication, an ICD, and lifestyle modifications. Additionally, these children's other parent has also been diagnosed with LQTS, but was identified as being affected after the children were diagnosed.

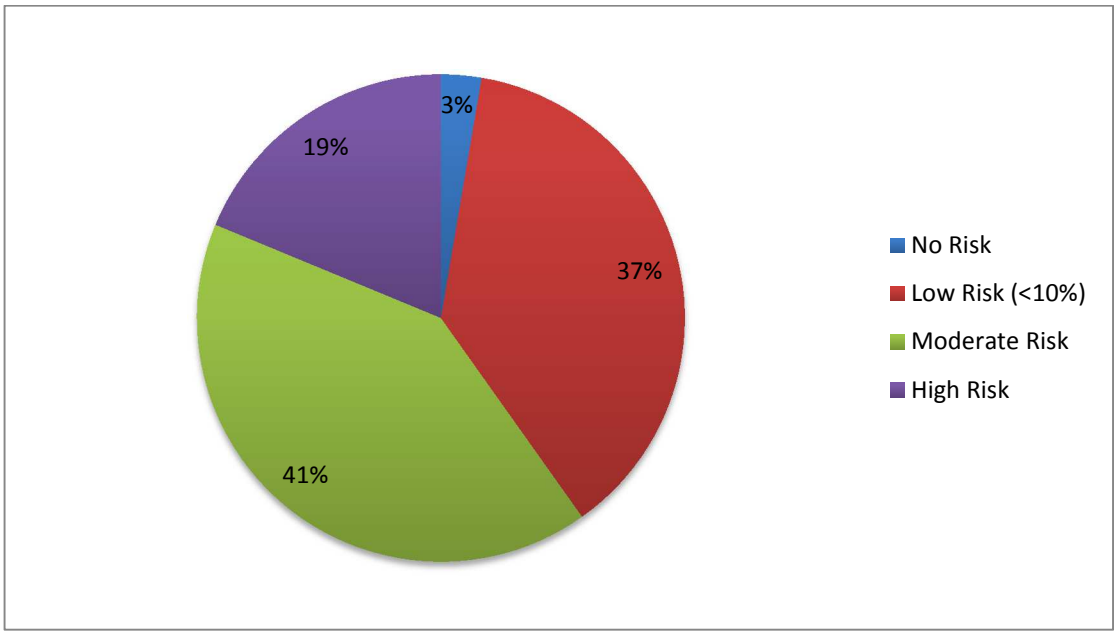

Figure 2.2 **Parental Understanding of the Risk of SADS for their Children**

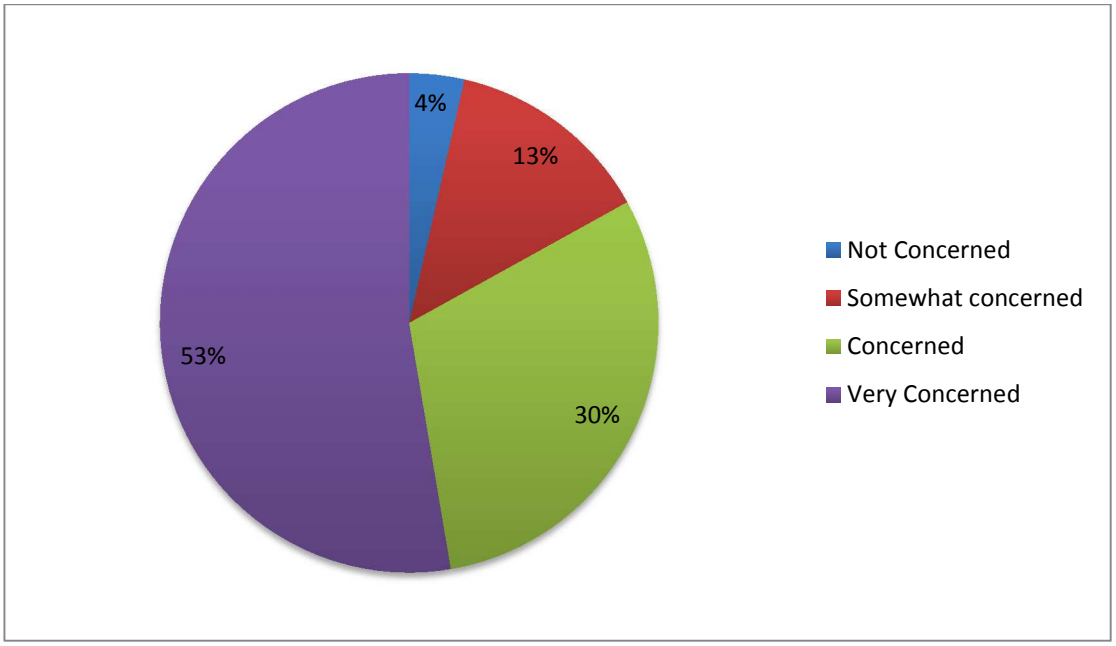

Figure 2.3 **Parental Level of Concern Regarding SADS**

Statistical analysis was conducted to investigate if the participants' understanding of the risk of SADS correlated with the symptoms the children have experienced or the parent's level of concern regarding SADS. Cochran-Mantel-Haenszel statistics revealed a correlation between the parents' understanding of their children's risk of SCA and whether or not their children have experienced arrhythmia ($p = .018$). Parents whose children have not experienced arrhythmia most often reported their children as having a low risk for SCA. In contrast, parents whose children have experienced arrhythmia most frequently stated that they felt that their children had a moderate risk of SCA. Utilization of the Monte Carlo estimate for Fisher's exact test identified a correlation between the parents' understanding of their children's risk of SCA and their level of concern regarding SCA ($p = .0021$). The greater the parents' perceived their children's risk of SCA to be, the more likely they were to report a higher level of concern about this risk.

Approximately 64 percent of respondents ($n = 69$) stated that they have specifically discussed the risk of sudden cardiac death with their children. When questioned about what prompted these conversations, the most commonly reported reasons were the diagnosis (61%) and healthcare appointments (49%). Both the child experiencing a symptom and a family member having an event prompted discussion about SADS approximately one-third of the time (Figure 2.3). About 23% of respondents listed other experiences, such as Facebook support groups or events surrounding medical management, which prompted discussion about the risk of SADS. Out of the 39 respondents who stated that they have not discussed this risk, the majority said it was because their children were too young and or they did not want to scare their children.

Table 2.5 Discussions Concerning the Risk of SADS

		Frequency	Percentage (%)
Have you discussed the risk of SADS with your child(ren)?		*n = 108*	
	Yes	69	64
	No	39	36
Events that prompted discussions about SADS		*n = 69*	
	Diagnosis	42	61
	Child experienced a symptom	23	33
	Family member had an event	22	32
	Other member of the community had an event	7	10
	A healthcare appointment	34	49
	Other	16	23

Out of the 69 respondents who have specifically discussed the risk of SCA with their children, 53 (77%) responded when asked about how they explained the risk of SCA to their children and what was most difficult about these conversations. Although the participants' answers were highly situation dependent, there were several responses that were repeatedly expressed. When explaining the risk of SCA to their children, participants most frequently focused on how the risk of SCA was the reason for the necessity of the treatment the children were undergoing. The possible symptoms and what actions to take if the children felt symptoms were also answers that were repeatedly reported. In addition, a number of parents used examples they felt their children could relate to, such as a professional athlete experiencing a SCA while being active or a relative who has previously experienced an event. Many parents also commented on the manner in which they discussed the risk of SADS. Phrases such as *"straight and to the point," "Blatantly. I didn't fluff it, "* and *"I told it like it is"* were commonly stated. Several parents also felt that being *"[honest] about the risks and diagnosis was the only*

way to go." Additionally, numerous participants reported explaining the information at a developmentally appropriate level.

Several themes were identified in responses to the question "What was the hardest aspect of these conversations for you as a parent?" The majority of parents felt that discussing the possibility of death and the emotional toll it has on both the parents and children was most difficult. Thoughts such as: *"I'm talking about my child dying. Every single day I worry it's his last. Every. Single. Day."* and *"seeing the fear in her eyes that she will die suddenly like her brother"* were commonly expressed. Many parents also felt that it was difficult to determine appropriate lifestyle modifications and enforce them. For example, one mother stated that "*seeing the pain in his eyes and feeling his heart break when his dream of college baseball was taken due to restriction in competitive sports*" was the hardest part of these conversations. Several parents also struggled with balancing the information they discussed with their children. In addition to *"keeping [the conversation] at a developmentally appropriate level,"* parents found it hard to inform their children about the seriousness of the condition without terrifying them. Lastly, a number of parents who also had a cardiomyopathy diagnosis reported struggling with feeling guilty for "*passing it on*" to their children.

2.4.6 Resources

Participants were also asked two questions regarding the use of resources when discussing their children's condition with them. Approximately 75% (n = 85) of participants responded when asked about what resources they found most helpful. The majority of respondents stated that they found healthcare professionals to be helpful in explaining the diagnosis. While physicians were the most commonly reported answer,

other professionals, such as genetic counselors, were also included. The Internet, specifically the SADS and HCMA websites, was the second most frequently stated resource that was thought to be helpful with the explanation. Participants also reported other affected people; books, visual aids, and pamphlets; and personal experience to be helpful with the explanation. A total of 71 participants responded when asked about what other resources would have been helpful to explain their children's diagnosis to them. The two most common responses were other affected people, specifically local support groups and peer groups for their children; and a counselor or psychologist to help cope with the diagnosis. Children-directed resources, such as storybooks or videos, were a third repeatedly expressed desire.

2.5 Discussion

Little research has been done to investigate how parents communicate with their children about their children's diagnosis of a cardiomyopathy associated with SADS. Based on data gathered from a survey completed by 114 parents with one or more children who have a diagnosis of a cardiomyopathy associated with SADS, this study was able to characterize the concerns parents had for their children related to their condition; the details of the initial explanation and follow-up conversations, such as who was involved, what was discussed, and the time frame in which it was discussed; and the aspects of these conversations that the parents found to be difficult. Furthermore, the responses from participants gave insight into the parents' perspective of their children's risk of SADS, how the parents discussed this risk with their children, and what aspects of these conversations they thought were most difficult. Lastly, the parents' statements

were informative about the resources that were both thought to be helpful and were desired, but not accessed.

2.5.1 Parental Concern

Three themes emerged when participants were asked what their biggest concern for their children regarding their condition. Most parents expressed concern about disease progression. If one or more of their children were asymptomatic, they were concerned about the development of symptoms. If their children were already symptomatic, their primary concerns were about experiencing SCA or requiring a transplant. A second theme was parental concern regarding their children's length and quality of life. Not only were parents worried about how long their children would live, but they also were concerned about how the number of healthcare appointments and lifestyle modifications would affect their daily life and emotional health. The last major identified theme was related to treatment; parents expressed concern about faulty treatments, treatment complications, and noncompliance by their children. While many of these concerns may be more difficult to address, parents in previous studies have made suggestions that have helped alleviate some of their concerns. These include having their children carry a cell phone, educating other caregivers and educators about the condition and associated risks, and teaching their children how to identify and avoid triggers (Farnsworth, 2006). Including recommendations such as these when disclosing children's diagnoses to their parents may help reduce the amount of anxiety they experience.

2.5.2 Communication Surrounding the Cardiomyopathy

Parents provided a variety of information regarding how the child's diagnosis was discussed within their family, including the details of the initial explanation as well as subsequent conversations about the condition. The children's mothers were significantly more likely than their fathers to be involved in the explanation of the diagnosis to their children. Although these results may in part be due to the fact that 86% of respondents were female, these findings are consistent with those found by D'Agincourt-Canning, who concluded that women are more likely to be responsible for communicating health-related information to other family members (2001). Interestingly, a correlation was found between the diagnosis and fathers' involvement in the explanation. Fathers whose children had a diagnosis of ion channelopathy were almost twice as likely to be involved in the explanation when compared to fathers whose children had a diagnosis of cardiomyopathy.

In agreement with recommendations made by multiple studies, children's diagnoses were most commonly explained to them gradually over a period of time (Mangset & Hofmann, 2014; Metcalfe et al., 2011; Anderson, 2008). A significant proportion of respondents stated that the initial explanation occurred over a period of days. Further investigation revealed that the initial explanation was more likely to occur over a shorter period of time as the age of diagnosis increased. This may be due to the fact that older children are more capable of understanding and digesting a greater amount of complicated information in a shorter period of time.

The most frequently discussed topics in both the initial and subsequent conversations were medical management and possible symptoms. Since the majority of

children were taking medication or have lifestyle modifications as a result of their condition, this is to be expected. Additionally, it makes sense for parents to want to explain what their children may experience. Two expected trends were found regarding the discussion of the genetic component of the condition with their children. A positive correlation was found between the age of diagnosis and the frequency at which genetics were discussed in the initial conversation; this is not surprising since more parents likely felt that their children could understand the information if they were older. Regardless of age at diagnosis, parents more frequently discussed genetics in subsequent conversations. Many parents may not have considered this information immediately relevant to their children. They also may have chosen to wait until their children are older because they felt their children were too young to understand such a complex concept. A significant correlation was also found between several topics discussed in follow-up conversations and whether or not their children were symptomatic. Possible symptoms, medical management, other family members with the condition, and how it specifically affects the heart were all reiterated more frequently during follow-up conversations in families whose children were symptomatic. This suggests that these aspects of the diagnosis may be less of a focus for the families whose children are asymptomatic.

The most commonly reported trigger for the conversations surrounded medical management, such as restrictions from physical activities, medications, and medical appointments. This is to be expected because these lifestyle changes and interactions with medical professionals have a daily impact on the lives of both the children and their parents. A significant correlation was identified between children's comments and questions being reported as a trigger and the specific diagnosis; parents whose children

have LQTS were over four times more likely to report children's comments and questions as a trigger compared to parents whose children have HCM. This may be due to the fact that children with HCM were twice as likely to be asymptomatic as children with LQTS; it would be expected for children to initiate conversation about their diagnosis more frequently if they were experiencing symptoms.

The most commonly reported aspects of these conversations that parents found difficult were related to medical management, the psychosocial and emotional implications of the condition, determining the appropriate amount and type of information, and possible symptoms, especially SCA. Not only did parents have difficulty enforcing the necessary limitations on physical activity, but many parents also reported uncertainty regarding which lifestyle modifications were truly necessary. Difficulty in determining appropriate lifestyle modifications was also reported in a study by Burns-Pentecost, who interviewed parents whose children had a recent diagnosis of LQTS (2013). Although not statically significant, a notable difference was found between the child's diagnosis and whether or not the parents found conversations surrounding medical management difficult. This difference may be due to the different treatments the children were undergoing and merits further investigation.

Participants also reported these conversations to be emotionally and psychologically difficult for both the parents and the children. The participants reported that both they and their children experienced fear regarding the children's physical health, as well as concern regarding quality of life and ability to cope with the implications of their condition. Specifically, some parents reported that their children struggled with "feeling different" and that they expressed desire to be more like their peers. Rahman reported

similar findings through interviews with children with an ICD and their parents (2013). In addition to feeling different than their peers, many children also expressed fear of the possibility of being shocked by their ICD. In contrast, only one participant reported being concerned about being different from his peers in a study done by Meulenkamp, who interviewed children who were carriers of LQTS, HCM, and familial hypercholesterolemia (2008). These conflicting results suggest that there may be additional psychosocial issues to address in the high-risk population being treated with an ICD compared to the patients who do not have an ICD or pacemaker.

Struggling with determining the appropriate way to convey information about the condition and the associated risks was a third theme that was identified in the participants' responses. In addition to making the information age-appropriate, parents were also unsure about how to explain the risks in a way that would not scare their children, but would make them understand the risks and be compliant with the recommended treatment. Several studies have underscored the importance of disclosing the information in an age-appropriate manner (Anderson, 2008; McConkie-Rosell, 2009). Being aware of the fact that parents are experiencing difficulty with conveying the diagnosis and associated risks in a manner that is developmentally appropriate and will instill appropriate concern will give healthcare providers the opportunity to proactively counsel parents on the disclosure process. Lastly, the fourth major recurring aspect that parents found difficult to discuss were the associated symptoms of the condition. Considering the serious of the potential symptoms, this is to be expected.

2.5.3 The Risk of SADS

In addition to exploring how a diagnosis of a cardiomyopathy or ion channelopathy associated with SADS is discussed in general, this study also investigated specifically how the risk of SADS is viewed and addressed. A positive correlation was found between the parents' understanding of their children's risk of SADS and their level of concern regarding this risk. This is to be expected; if parents think their children have a higher risk for SADS, they are more likely to be more concerned about this risk. Even though this association was identified, it is important to note that while most parents viewed their children's risk for SADS as either low or moderate, the majority of parents were still very concerned about this risk. These results agree with the data obtained by Hendricks et al., who found that most parents of children with LQTS were highly concerned about the possibility of sudden death (2005).

The majority of participants had previously discussed the risk of SADS with their children. Several triggers were reported when asked what prompted these conversations; the more common being the diagnosis itself and healthcare appointments. Other triggers, such as the child or someone else experiencing a symptom, Facebook support groups, and events related to medical management were also reported. The most common reasons for not discussing this risk were that their children were too young or that the parents did not want to scare them. These explanations are similar to the responses given by parents of children with LQTS and HCM in the study conducted by Meulenkamp (2008), who also found that a minority of parents chose to limit the amount of information they revealed to their children.

Parents who had discussed the risk of SCA with their children were asked about how exactly it was communicated. Framing the risk of SADS as the reason for treatment, the possible symptoms, and what steps need to be taken if the children experience symptoms were the most commonly discussed topics. Several parents who related the risk of SADS to the importance of treatment commented that this approach was their way of encouraging compliance with the recommended lifestyle modifications and pharmaceutical interventions. This approach may be helpful for parents whose children are resisting the recommended treatment plan and are old enough to comprehend the connection. The participants who commented on the manner in which they described the risk of SADS stated that they explained the risk directly and honestly. These responses are in accordance with previous studies that investigated how children prefer to learn about the risks associated with a genetic condition (Rowland & Metcalfe, 2013; Meulencamp, 2008). These investigators concluded that a straightforward explanation helped both the children and the parents cope with the diagnosis.

There was overlap in the responses when parents were asked what they found to be the most difficult about these conversations compared to discussion about the diagnosis in general. When specifically discussing the risk of SCA, most people stated that they found discussing death and emotional implications it has on the family most difficult. Considering that many parents report experiencing fear regarding their children's risk of SCA, this was expected (Farnsworth, 2006). Parents found it difficult to determine the correct balance of information to share with their children during these conversations as well. An emotion that parents struggled with during conversations about the risk of SCA but did not express in responses to the previous question was guilt

for passing the condition on to their children. Feelings of guilt have been identified in affected parents with children who had a LTQS diagnosis in previous studies (Burns-Pentecost, 2013). Heightened awareness of the possibility of parental guilt in the healthcare community may increase the likelihood of providers proactively addressing it at the time of a child's diagnosis.

2.5.4 Resources

This study asked participants what resources they found to be helpful when communicating their children's diagnosis to them as well as what additional resources they would have liked. Cardiologists, websites of organizations such as HCMA and SADS, other affected people, books and pamphlets, and personal experience were all reported to be helpful for discussing the diagnosis with their children. The most commonly requested resources were related to helping their children better understand and cope with the diagnosis and included: local or peer support groups for their children, a counselor or psychologist to help cope with the diagnosis, and child-oriented pamphlets, storybooks, and videos. Previous research has identified online peer support groups as a resource that may aid in the coping process (Burns-Pentecost, 2013). In addition, a study conducted by Conlin determined that while many pediatric cardiologists are screening for psychosocial stressors, most do not make referrals to psychological services on a regular basis (2012). However, the participants did state that they would be more likely to make a referral if resources were easily accessible. Together, these findings suggest that establishing stronger relationships between pediatric cardiologists and psychological services may increase the referral rate and help address the lack of

psychological support available to aid in coping with the implications of a cardiomyopathy associated with SADS.

2.5.5 Limitations and Future Research

Common limitations of Internet-based research, such as the self-selected sample, unknown response rates, and inability to control who accessed the survey are all limitations of this study. The individuals who chose to participate in the study were likely the most active members of the ion channelopathy and cardiomyopathy organizations and Facebook support groups. Participants were also invited to participate in the survey through several diagnosis-specific Facebook groups and organizations; this method of obtaining participants may have biased the participant population. The homogeneity of the respondents must also be noted; most individuals reported being Caucasian and female. Therefore, the data collected may not be an accurate representation of the actions and opinions of the general population.

Future studies on this topic that include a more heterogeneous participant pool may produce less biased data. Additional research could also investigate which aspects of these conversations went well and went poorly as well as obtain recommendations from parents on the best way to discuss children's diagnoses with them. This data could then be provided to parents of children who have recently been diagnosed with a cardiomyopathy associated with SADS. Finally, researchers could obtain the perspective of individuals who were diagnosed as children to gain their input on what they viewed as helpful and what resources they would have wanted as they learned about their diagnosis.

2.6 Conclusions

This study was conducted to gain a better understanding of how a child's diagnosis of a heart condition associated with SADS is discussed within families. Participants were asked about the details of both the initial explanation and later conversations as well as how the risk of SCA was viewed and addressed. The majority of participants thought their children either had a moderate (41%) or low (37%) risk for SCA and were highly concerned (53%) about this risk. Parents reported explaining the risk of SCA in straightforward manner; this approach is thought to help the children and their parents cope with the risk. A number of participants also related the risk of sudden death to why the prescribed treatment was necessary. They felt that this line of reasoning would encourage their children to be more compliant with the recommended medication regimen and lifestyle modifications.

Information was also gathered on what aspects of these conversations parents found to be difficult. Participants reported struggling with determining and enforcing lifestyle modifications, managing their own as well as their children's emotional reactions, determining the amount and type of information to share with their children, and discussing the risk of the more serious symptoms. Several parents also reported feeling guilty for passing the condition onto their children. Concerns regarding disease progression, length and quality of life, and treatment failures and complications were expressed as well.

Finally, parents provided feedback on what resources were helpful and what resources they desired. Healthcare professionals, the Internet, other affected people, visual aids, and personal experience were all thought to be useful for discussing the

SADS condition with their children. Services and resources that were most frequently requested were support groups for their children, a counselor or psychologist, and child-oriented materials.

Increased awareness of how families communicate about children's diagnosis of a SADS condition, including the aspects parents struggle with, as well as parents' main concerns and what resources are needed will allow healthcare providers to be more proactive about addressing these issues from the time of the initial diagnosis. It is thought that this intervention will in turn facilitate more open, age appropriate discussion of the condition and help children and their parents more successfully cope with the implications of the diagnosis.

Chapter 3. Conclusions

This study was conducted to gain a better understanding of how a child's diagnosis of a heart condition associated with SADS is discussed within families. Participants were asked about the details of both the initial explanation and later conversations as well as how the risk of SCA was viewed and addressed. The majority of participants thought their children either had a moderate (41%) or low (37%) risk for SCA and were highly concerned (53%) about this risk. Parents reported explaining the risk of SCA in straightforward manner; this approach is thought to help the children and their parents cope with the risk. A number of participants also related the risk of sudden death to why the prescribed treatment was necessary. They felt that this line of reasoning would encourage their children to be more compliant with the recommended medication regimen and lifestyle modifications.

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Appendix A- Request to Host Survey

To Whom it May Concern

My name is Kristin Wiley and I am a graduate student in the Genetic Counseling Program at the University of South Carolina School of Medicine. For my thesis project I plan to gain a better understanding of how parents communicate with their children about a diagnosis of a cardiac condition associated with sudden arrhythmic death by surveying parents that have or have had a child with one of these conditions. My survey, which is attached, inquires first about how parents explained their child(ren)'s diagnosis to them and then specifically asks questions about how they have discussed the risk of sudden arrhythmic arrest/death. I realize that this is a sensitive subject for many families; however, I believe that by learning more about how this information is communicated within families, it will help guide practitioners in addressing these issues with parents in the future. I would greatly appreciate your help in distributing my survey to eligible parents. The more participants we have, the more we can learn about how to help them. If you are willing to pass along my survey to your members please let me know. You can contact me at kristinannewiley@gmail.com.

Best,

Kristin Wiley

Appendix B – List of Social Media Sites Where Survey Links Were Posted

1. Long QT Syndrome Support and Learning Community:
   https://www.facebook.com/groups/lqtssupportandlearningcommunity/
2. LQTS Kids & Families - for anyone affected by Long QT Syndrome:
   https://www.facebook.com/groups/193978400681765/
3. Cardiac Arrythmia Support Group:
   https://www.facebook.com/groups/15750922307/
4. Hypertrophic Cardiomyopathy....GROUP:
   https://www.facebook.com/groups/418243221551082/#_=_
5. SADS- Sudden Arrhythmia Death Syndrome:
   https://www.facebook.com/groups/191810694204543/
6. Brugada Syndrome Awareness:
   https://www.facebook.com/groups/206381569393542/
7. Dilated Cardiomyopathy: https://www.facebook.com/groups/8611170489/
8. Children's Cardiomyopathy Foundation:
   https://www.facebook.com/pages/Childrens-Cardiomyopathy-Foundation/75335952379
9. SADS (Sudden Arrhythmia Death Syndromes) Foundation:
   https://www.facebook.com/pages/SADS-Sudden-Arrhythmia-Death-Syndromes-Foundation/19466599975

Appendix C- Invitational Letter to Participate in Survey

Dear Potential Participant,

You are invited to take part in a graduate student research study looking into how parents communicate with their children about the children's diagnoses of cardiac diseases associated with sudden arrhythmic death syndrome (SADS). Participation involves completing an online questionnaire about your child's diagnosis, your communication with your child(ren) about different aspects of the condition, and the resources you used during those conversations.

All survey responses will be kept anonymous and confidential. The data collected during this study may be published or presented at medical conferences, but your responses will not be associated with any personally identifying information. The survey will take 15-20 minutes to complete. Your participation in this research study is voluntary. If you come across a question that you do not wish to answer, please skip it and continue with the survey. You may also choose to not complete the survey at any time. By completing the survey, you are consenting that you have read and understand this information.

As a thank you for participating in our study, you may choose to enter into a drawing to win a $25 gift card to the store or restaurant of your choice. You do not have to complete the survey to be eligible for the drawing. If you would like to enter into the drawing, please enter your contact information in the boxes provided at the bottom of the survey. Your contact information will only be used to send the winner his/her gift certificate and will not be associated with your responses. The winner will be drawn after the study is complete.

Thank you for taking the time to participate in this study. Your answers will help healthcare professionals such as cardiologists and genetic counselors address common obstacles present at the time of diagnosis and aid parents in communicating with their children about their condition. If you have any questions about this research study, please contact me or my advisor Erin Demo at the information below. If you have any questions about your rights as a research member, you may contact the Office of Research Compliance at the University of South Carolina at (803)777-7095.

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Appendix D – Online Survey

SECTION 1: YOUR CHILDREN'S DIAGNOSIS

1. How many of your children have been diagnosed with a cardiac condition associated with SADS?
    - One
    - Two
    - Three or more

2. What is the gender of your child(ren) who has/have been diagnosed

	Child 1	Child 2	Child 3
Female			
Male			
N/A			

3. What is your child(ren)'s diagnosis?
    - Long QT syndrome (LQTS)
    - Hypertrophic cardiomyopathy
    - Brugada syndrome
    - CPVT (catecholaminergic polymorphic ventricular tachycardia)
    - Arrhythmic right ventricular cardiomyopathy (ARCV)
    - Dilated Cardiomyopathy (DCM)
    - Other (please specify)

4. At what age was your child diagnosed?

Age	Child 1	Child 2	Child 3
0-3			
4-6			
7-9			
10-12			
13-15			
16-18			
19-21			
22+			
N/A			

5. Is/are your child(ren) alive or deceased

Status	Child 1	Child 2	Child 3
Alive			
Deceased			

6. How old is/are your child(ren) now?

Age	Child 1	Child 2	Child 3
0-3			
4-6			
7-9			
10-12			
13-15			
16-18			
19-21			
22+			
N/A			

7. Have either you or your child's other parent been diagnosed with the same SADS condition? If so, who has been diagnosed?
    - Yourself
    - Your child's other parent
    - Neither parent has been diagnosed

8. Who was the first person to be diagnosed in the family?
    - One of your children
    - Mother of the child(ren)
    - Father of the child(ren)
    - Grandparent of the child(ren)
    - Other (please specify)

9. What was/were your child(ren)'s first presenting symptom?

Symptom	Child 1	Child 2	Child 3
Syncope (fainting)			
Arrhythmia (abnormal heart rhythm)			
Sudden death			
Seizure			
They have not had any symptoms yet			
N/A			
Other (please specifiy)			

10. How was/were your child(ren) diagnosed? (Please select all that apply)

Test method	Child 1	Child 2	Child 3
Electrocardiogram (EKG/ECG)			
Echocardiogram			
Genetic Testing			
Autopsy			
Physical exam			
Exercise test			
Holter monitor (24 hr EKG)			
Other (please specify)			

11. What symptoms has/have your child(ren) experienced to date? (Please select all that apply)

Symptom	Child 1	Child 2	Child 3
Syncope (fainting)			
Arrhythmia (abnormal heart rhythm)			
Sudden death			
Seizure			
Sudden cardiac arrest			
Dizziness			
Other (please specify)			

12. What treatments is your child(ren) currently receiving for this condition? (Please select all that apply)

Treatment	Child 1	Child 2	Child 3
Medication (like beta-blockers)			
ICD: Implantable Cardioverter Defibrillator)			
Pacemaker			
Lifestyle modifications (like restriction from competitive sports)			
Other (please specify)			

13. What s your biggest concern for your child(ren) regarding their condition?

SECTION 2: RISK OF SADS

The following questions will ask you about your outlook regarding your child(ren)'s risk of SADS

14. What do you understand to be your child(ren)'s risk of sudden cardiac arrest?
    - No risk
    - Low risk (<10%)
    - Moderate Risk
    - High Risk

15. How concerned are you about your child(ren)'s risk of SADS
    - Not concerned
    - Somewhat concerned
    - Concerned
    - Very concerned

SECTION 3: COMMUNICATION WITH YOUR CHILD

The following questions will ask you about how you communication with your child about their diagnosis

16. Who was involved in explaining your child's diagnosis to him/her? (Please select all that apply)

	Child 1	Child 2	Child 3
Mother			
Father			
Other family member			
Physician			
Genetic Counselor			
Other (please specify)			

17. Over what period of time did the explanation of their diagnosis take place?

Timer Period	Child 1	Child 2	Child 3
Days			
Weeks			
Months			
Years			

18. What was the primary focus of your initial explanation of his/her condition? (Please select all that apply)

	Child 1	Child 2	Child 3
Genetic aspect of the condition (such as the inheritance apptern, chance of passing it down to their childred)			
Possible symptoms (arrhythmia, syncope, seizures, sudden death, dizziness)			
Other family members with the same condition			
Medical Management (medications, ICD implant, lifestyle modifications (like limitations on physical activity))			
How the condition specifically affects the heart			
Other (please specify)			

19. What topics were focused on during subsequent conversations? (Please select all that apply)

	Child 1	Child 2	Child 3
Genetic aspect of the condition (such as the inheritance apptern, chance of passing it down to their childred)			
Possible symptoms (arrhythmia, syncope, seizures, sudden death, dizziness)			
Other family members with the same condition			
Medical Management (medications, ICD implant, lifestyle modifications (like limitations on physical activity))			
How the condition specifically affects the heart			
Other (please specify)			

20. Is your child(ren)'s condition a regular topic of conversation? If so, how often is it discussed?

Time Period	Child 1	Child 2	Child 3
Daily			
Weekly			
Monthly			
It is not discussed regularly			

21. What triggers these conversations?

22. What were the more difficult aspects of these conversations with your child? Some examples include: explaining the biology, discussing lifestyle modifications, describing associated risks, and managing your emotions.

SECTION 4: CONVERSATION ABOUT SADS

23. Have you ever specifically discussed your child(ren)'s risk of sudden cardiac arrest with them? If not, why?
    - Yes
    - No

These questions will ask you about the conversations you have had with your child(ren) regarding their risk of SADS

24. What has prompted these conversations? (Please select all that apply)
    - Diagnosis
    - Child experienced a symptom
    - Family member had an event
    - Other member of your community has an event
    - A healthcare appointment
    - Other (please specify)

25. How exactly did you explain this risk of sudden cardiac arrest to your child(ren)? (ex: Who was involved? How did you phrase it? What did you emphasize?)

26. What was the hardest aspect of these conversations for you as a parent?

SECTION 5: RESOURCES

The following questions will ask you about the resources you used to help communicate your child(ren)'s diagnosis to them.

27. What resources did you find most helpful in talking to your child(ren) about his/her diagnosis? Some examples include pamplets, health professionals, the internet, etc.

28. Looking back, what other guidance or resources would you have wanted to help talk with your child(ren) about their condition?

Thank you for sharing your family's experience with a SADS condition. The following questions will ask about basic demographic information, such as your age and gender.

SECTION 6: DEMOGRAPHICS

29. What is your age?
    - <20
    - 20-29
    - 30-39
    - 40-49
    - 50-59
    - 60-69
    - >70

30. What is your gender?
    - Female
    - Male

31. What is the highest level of school you have completed or the highest degree you have received?
    - Less than high school degree
    - High school degree or equivalent (e.g. GED)
    - Some college but no degree
    - Associate degree
    - Bachelor degree
    - Graduate degree

32. What is your ethnicity? (Please select all that apply)
    - American Indian or Alaskan Native
    - Asian or Pacific Islander
    - Black or African American
    - Hispanic or Latino
    - White/Caucasian
    - Prefer not to answer

33. Which of the following best describes your current relationship status?
    - Married
    - Widowed
    - Divorced
    - Separated
    - In a domestic partnership or civil union
    - Single, but cohabitating with a significant other
    - Single, never married

If you wish to enter the drawing for the $25 gift card to the store or restaurant of your choice, please enter your contact information below. This information will only be used in the drawing and will not be connected to your answers.

34. Contact Information
    - Name:
    - Street Address:
    - City:
    - State:
    - Zip Code:
    - Telephone Number:
    - Email Address:

Appendix E – Additional Data

		Frequency	Percentage (%)
Number of Affected Children in the family		*N=114*	
	One	79	69.3
	Two	29	25.4
	Three	6	5.3
Parents' Diagnosis		*N=112*	
	Participant is affected	54	48.2
	Child(ren)'s other parent is affected	16	14.3
	Neither parent is affected	42	37.5
First Person in the Family to be Diagnosed		*N=114*	
	Child	61	53.5
	Mother of child	26	22.8
	Father of child	5	4.4
	Grandparent of child	7	6.1
	Great-grandparent of child	4	3.5
	Aunt or uncle of child	5	4.4
	Other	6	5.3

		Frequency			Percentage (%)		
		Child 1 *N=114*	Child 2 *N=35*	Child 3 *N=6*	Child 1	Child 2	Child 3
Vital Status							
	Alive	108	34	6	94.7	97.1	100
	Deceased	6	1	0	5.3	2.9	0
Method of Diagnosis							
	EKG	71	18	6	64.0	51.4	100
	ECHO	55	14	5	48.2	40.0	83.3
	Genetic Testing	54	22	6	47.4	62.9	100
	Autopsy	3	0	0	2.6	0	0
	Physical Exam	20	4	3	17.5	11.4	50
	Exercise Test	30	9	4	26.3	25.7	66.7
	Holter	38	12	5	33.3	34.3	83.3
	Other	9	---	---	7.9	---	---

		Frequency		Percentage (%)	
		Child 2	Child 3	Child 2	Child 3
Gender		*N=35*	*N=6*		
	Female	16	4	45.7	66.7
	Male	19	2	54.3	33.3
Age at Diagnosis		*N=35*	*N=6*		
	0-3	6	3	17.1	50.0
	4-6	5	1	14.3	16.7
	7-9	9	2	25.7	33.3
	10-12	7	0	20.0	0
	13-15	4	0	11.4	0
	16-18	3	0	8.6	0
	19-21	1	0	2.9	0
	22+	0	0	0	0
Current Age		*N=35*	*N=6*		
	0-3	0	0	0	0
	4-6	2	1	5.7	16.7
	7-9	8	4	22.9	66.7
	10-12	13	1	37.1	16.7
	13-15	3	0	8.6	0
	16-18	2	0	5.7	0
	19-21	2	0	5.7	0
	22+	4	0	11.4	0
	Deceased	1	0	2.9	0
First Presenting Symptom		*N=30*	*N=6*		
	Syncope	5	0	14.3	0
	Arrhythmia	3	2	8.6	33.3
	Sudden Death	1	0	2.9	0
	Seizure	0	0	0	0
	Heart murmur	0	0	0	0
	Shortness of breath	0	0	0	0
	Asymptomatic	19	4	54.3	66.7
	Other	3	0	8.6	0

		Frequency		Percentage	
		Child 2 *N=35*	Child 3 *N=6*	Child 2	Child 3
People Involved in the Explanation					
	Mother	30	6	85.7	100.0
	Father	17	5	48.6	83.3
	Other Relative	2	1	5.7	16.7
	Physician	24	6	68.6	100.0
	Genetic Counselor	7	2	20.0	33.3
Time Frame for the Explanation					
	Days	12	3	34.3	50.0
	Weeks	6	0	17.1	0
	Months	7	1	20.0	16.7
	Years	10	2	28.6	
Primary Focus of the Initial Explanation					
	Genetics	13	2	37.1	33.3
	Possible Symptoms	18	5	51.4	83.3
	Other affected family members	16	3	45.7	50.0
	Medical Management	16	6	45.7	100.0
	How the condition specifically affects the heart	15	6	42.9	100.0
Topics discussed during subsequent explanations					
	Genetics	18	4	51.4	66.7
	Possible Symptoms	21	6	60.0	100.0
	Other affected family members	23	6	65.7	100.0
	Medical Management	17	4	48.6	66.7
	How the condition specifically affects the heart	15	6	42.9	100.0
Frequency of discussion					
	Daily	6	0	17.1	0
	Weekly	12	4	34.3	
	Monthly	5	1	14.3	16.7
	Not discussed regularly	10	1	28.6	16.7