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Exercise Recommendations for Active Adults at Risk for Sudden Cardiac Death: “Can I Continue to Exercise?”

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**Exercise Recommendations for Active Adults at Risk for Sudden Cardiac Death:
“Can I Continue to Exercise?”**

by

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Submitted in Partial Fulfillment of the Requirements

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Abstract

Sudden arrhythmic death syndrome (SADS) is due to genetic conditions associated with cardiac ion channelopathies and cardiomyopathies. Many of these conditions have exercise guidelines regarding eligibility and disqualification recommendations for competitive athletes through the American Heart Association and the American College for Cardiology. This study investigates how medical professionals communicate these exercise recommendations to patients, and in turn, how well patients understand these recommendations. The study also explores motivations for either continuing or discontinuing an exercise practice after a diagnosis. A questionnaire was distributed through cardiac disease-focused support groups and organizations. Data from 67 individuals with a SADS condition were analyzed for possible associations and significant differences. Results demonstrated that intensity of exercise on a 1 to 9 scale decreased from 4.9 to 3.3 after a diagnosis ($p < .01$). Time spent exercising decreased from 204 minutes per week to 142 min/week after a diagnosis ($p < .01$). Patients were significantly more likely to decrease activity intensity when they felt confident with the communication of their exercise recommendations and how well they understood them. This study showed that individuals felt poorly informed regarding their exercise recommendations and often take on heavy emotional burdens due to restrictive guidelines. The respondents widely expressed that quality of life is an important factor in their decisions about exercise, and that continuing exercise is a major component of a

healthy lifestyle despite risks associated with SADS. Involving genetic counselors in the discussion of exercise guidelines is recommended while addressing the psychosocial outcomes of a recent diagnosis. This addition to the management of individuals with a SADS diagnosis is likely to increase awareness of how exercise recommendations are managed and how well patients understand that their personalized exercise plan will promote a safer healthcare management plan for each patient.

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Chapter 1: Background

1.1 Overview of Electrophysiologic and Structural Congenital Heart Disease

Congenital heart disease (CHD) is the most common birth defect and is defined as a number of different problems affecting the heart at the time of birth. Although congenital, some of these problems may not show physical presentations until years later. Sudden Cardiac Death (SCD), causing an estimated 325,000 adult deaths in the United States each year, is defined as an unexplained death caused by loss of heart function (Napolitano, Bloise, Monteforte, & Priori, 2012). Some genetic cardiovascular diseases can result in SCD and include but are not limited to cardiomyopathies and channelopathies. Specifically, each diagnosis has its own etiology, incidence, genetic cause, and risk of SCD (Sheikh et al., 2015). Research on these congenital heart diseases has primarily focused on young athletes to better determine the epidemiology of each individual classification of disease.

Cardiomyopathies refer to diseases of the heart muscle, where the muscle tissue becomes enlarged, thick, or rigid. In rare and severe cases, the muscle tissue is replaced, making the heart weaker and prone to arrhythmias. Hypertrophic cardiomyopathy (HCM) is typically an inherited genetic condition that leads to a thickened heart muscle around the left ventricle, making it rigid and dysfunctional. Specifically, thickening occurs in the interventricular septum, which separates the right and left ventricle. In severe cases, the septum can obstruct the flow of oxygen-rich blood from exiting the heart. This can lead to symptoms including fatigue, syncope, and feeling out of breath. Sometimes the

thickening and scarring of the heart muscle changes the integrity of the communication between cells, causing arrhythmias (National Library of Medicine, 2016). These arrhythmias and possible SCD are often more likely to occur during intense physical activity (Sheikh et al., 2015).

The prevalence of HCM is assumed to be 1 in 500, and is known to be the leading identified cause of SCD in competitive athletes (Maron, Haas, Ahluwalia, Murphy, & Garberich, 2016b). As the most common genetic heart disease in the United States, cardiovascular physicians and genetic counselors recognize the variable expressivity of the condition. Age of onset can range from childhood to late adulthood, with some individuals never showing signs or symptoms at all. Dozens of genes can cause hypertrophic cardiomyopathy, with the most common being *MYH7*, *MYBPC3*, *TTNT2*, and *TNNI3*. All of these genes play important roles in forming sarcomeres, which are the basic units of muscle contraction. Hypertrophic cardiomyopathy is a perfect example of genetic heterogeneity, and each individual case is unique (Maron et al., 2015).

Another common genetic cardiomyopathy that can cause SCD is arrhythmogenic right ventricular cardiomyopathy (ARVC). In ARVC, the proteins holding the heart muscle cells together do not develop properly, leading to fatty deposits that build up in an attempt to repair the damage. This usually affects the right side of the heart and can cause arrhythmias, which can lead to SCD, especially during strenuous exercise (Napolitano et al., 2012).

The frequency of ARVC is estimated to be 1 in 1,000 to 1 in 1,250, but may be under-diagnosed due to the difficulty in diagnosing the condition. Unfortunately, most affected individuals do not show signs or symptoms until a severe cardiac event happens,

which is typically in adulthood. The most common genetic changes are in the *PKP2* gene, which is involved in the function of desmosomes. Desmosomes act as the cell-to-cell “superglue” that play a role in signaling between neighboring cells and give strength to the heart muscle itself (National Library of Medicine, 2016).

Channelopathies are diseases that result from the dysfunction of ion channels located in the membranes of cells, and specifically in cardiac tissue. These diseases can lead to irregular heart rhythms, syncope, arrhythmias, and SCD. A common example of a channelopathy is long QT syndrome (LQTS). LQTS is an arrhythmogenic disorder that presents with a prolonged QT interval (> 440 milliseconds in males and > 460 ms in females). LQTS exists with a variety of genotypes, meaning that the condition may be caused by a deleterious mutation in one of several genes. Each unique genotype is typically associated with a different complex phenotype, differing arrhythmogenic triggers, and mutations in various genes (Basavarajaiah et al., 2007). LQTS1 is associated with cardiac events during exercise, LQTS2 events are triggered by alarm clocks or moments of surprise, and events in LQTS3 typically occur during sleep. These events can occur from infancy to adulthood (National Library of Medicine, 2016).

The prevalence of all LQTS types is estimated to be 1 in 2,000. Genetic changes in genes that provide instructions for sodium and potassium channels cause LQTS, and the most common genes are *KCNE1*, *KCNE2*, and *SCN5A*. With no structural changes in the heart, the key diagnostic tool is the ECG (Electrocardiogram), which typically shows the prolongation of the QT interval. In some cases, individuals may be “gene-positive, phenotype-negative,” meaning these individuals carry a known gene mutation but do not show signs or symptoms of the condition (Basavarajaiah et al., 2007).

In addition, Short QT (SQT) syndrome also affects the QT interval and can be associated with arrhythmias and SCD. Symptoms including dizziness, syncope, and/or cardiac arrest can occur anytime from early infancy to old age. Genetic changes in potassium channel genes, *KCNH2*, *KCNJ2*, and *KCNQ1*, cause SQTS. Much less is known about this condition, as only a few hundred individuals have been identified worldwide (Napolitano et al., 2012).

Another channelopathy, Brugada syndrome, affects the ST segments on the patient's ECG. This condition leads to ventricular arrhythmias, and if left untreated can cause fainting, seizures, difficulty breathing, and sudden cardiac death. Cardiac events most often occur during rest or sleep, but are also known to occur during exercise (Napolitano et al., 2012). Signs and symptoms of Brugada syndrome typically become apparent in adulthood, but can occur from early infancy to late adulthood. This condition may explain some cases of SIDS, and cases of sudden death before the age of 40 years (National Library of Medicine, 2016).

The frequency of Brugada Syndrome is estimated to affect 1 in 2,000 individuals worldwide, with a higher prevalence in Asian ancestries. Brugada Syndrome is also eight to ten times more common in men than in women due to the synergistic effects of testosterone. The most commonly mutated gene in Brugada Syndrome is *SCN5A*, which reduces the flow of sodium ions into heart muscle cells (National Library of Medicine, 2016). This condition is typically treated with strict exercise guidelines regarding disqualification from competitive sports that may lead to traumatic injury or loss of consciousness due to the higher likelihood of cardiac event during sleep, rest, or loss of consciousness (Maron et al., 2015).

Other conditions that may cause SCD include but are not limited to familial dilated cardiomyopathy (DCM), catecholaminergic polymorphic ventricular tachycardia (CPVT), and left ventricular non-compaction cardiomyopathy (LVNC). These cardiovascular syndromes are all inherited conditions that are associated with considerable risk of cardiac events (Maron et al., 2015). A comprehensive table describing the genetics, inheritance pattern, and prevalence of these conditions can be found in Appendix E.

1.2 Genetics of Sudden Cardiac Death

Genetic testing for these congenital heart diseases has improved dramatically in the past five to ten years, and cardiac multigene panel testing is now widely available for symptomatic patients. Family members of a patient with a history of SCD or with significant symptoms are encouraged to seek genetic counseling for potential genetic testing when a known mutation has been identified in one close family member (Ormondroyd, Oates, Parker, Blaire, & Watkins, 2014). Approximately 5% to 15% of cardiac arrest victims fail to show evidence of structural abnormalities at the time of autopsy. If the individual is then genetically diagnosed at autopsy with idiopathic left ventricular fibrillation, recommendations for testing for the specific mutation are communicated to close family members. This fact, in addition to incomplete penetrance and variable expressivity of the various genotypes, highlights the importance of genetic testing for all family members at risk (Napolitano et al., 2012).

Penetrance is described as the proportion of people with a particular mutation who exhibit signs and symptoms of a genetic disorder. Variable expressivity refers to the wide range of signs and symptoms that can manifest in different individuals with the same

genetic condition. Variable expressivity and reduced penetrance result from a combination of environmental, genetic, and lifestyle factors. Most of the cardiomyopathies and channelopathies previously described are known to show variable expressivity and reduced penetrance, with some family members with the mutation showing minimal or no symptoms. This often presents as a challenging case with seemingly no suggestive family history. Thus, genetic counselors offer a closely targeted family history intake, with focused questions about a known set of possible phenotypic symptoms within a three-generation family history.

Families with inherited cardiac conditions will have affected individuals who showed signs or symptoms at completely different ages. Symptoms may also vary among family members with the same mutation including chest pain, shortness of breath, syncope, palpitations, cardiac arrest, and those reporting with no symptoms. This is especially seen in families with HCM, making it difficult to predict genetic testing outcomes for family members (Cirino & Ho, 2014). Since some affected individuals show no symptoms, genetic testing of unaffected close family members is a critical step in making a genetic diagnosis when a deleterious mutation has been identified in a clinically affected individual.

ARVC is an autosomal dominant condition with age-related penetrance and significant variable expressivity. This cardiomyopathy in addition to HCM varies in the age of onset and clinical presentation for each individual, sometimes even with the same familial mutation. A review from 2015 states that exercise plays a major role in disease penetrance and risk for SCD in ARVC (James, 2015). When analyzing a possible inheritance pattern within a three-generation family history, the age of onset is critical to

assess for possible physical manifestation and cardiac events in family members. This challenging task is typically performed by a genetic counselor.

Similarly, another publication by (Earle et al., 2014) demonstrated that small changes in arrhythmia genes modify the risk of cardiac events and SCD in long QT syndrome. They identified two small changes (single base DNA misspellings called single nucleotide polymorphisms, or SNPs) in both of the *NOS1AP* and *KCNQ1* genes that are associated with an increased risk for arrhythmia. Several other SNPs were suggested to show both positive and negative associations with QT length on an ECG (Earle et al., 2014). Even the smallest changes in arrhythmia genes can have critical phenotypic effects on patients, stressing the importance of personalized genetic counseling for each individual family and each genetically diagnosed patient based on their unique genetic change.

Counseling families effectively on the unique genetic cause of cardiovascular disease in their family, the risk of SCD or other complications, specific triggering events, and medical management is challenging. Therefore, appropriate knowledge in genetics and the ability to communicate that information to patients in an understandable manner is critical to provide relevant healthcare for each individual. Appropriate management of each case is dependent on the specific genetic cause of the specific cardiovascular condition, as well as the environmental factors that could contribute to that individual's risk.

1.3 The Bethesda Guidelines and Current AHA/ACC Exercise Recommendations

Despite the health benefits that typically result from regular exercise in most individuals, the small subset of individuals who are at risk for SCD must weigh the

possible risks that come with exercise. Many individuals diagnosed with HCM, ARVC, LQTS types 1 and 3, SQT Syndrome, Brugada syndrome, and other similar diagnoses are at an elevated risk for arrhythmias, syncope, and SCD at times of strenuous physical activity (Sheikh et al., 2015; Napolitano et al., 2012). A significant amount of research has been reported on SCD in competitive athletes, and studies have determined that sudden arrhythmic death syndrome (SADS) was the most prevalent cause of death at young ages in these athletes. Specifically, a study performed between 1994 and 2014 of 357 young competitive athletes who died suddenly showed that 42% of the cases were due to SADS: 13% due to ARVC; 6% due to HCM; and 13% due to idiopathic left ventricular hypertrophy (Finocchiaro et al., 2016). In another study performed on 842 young competitive athletes with autopsy-confirmed cardiovascular diagnoses through the U.S. National Registry of Sudden Death in Athletes, HCM was the most common diagnosis, occurring in 36% of patients; with ARVC diagnosed in 13%; and LQTS diagnosed in 7% of the athletes (Maron et al., 2016b).

With recent research and new knowledge about the gene-specific etiologies of hereditary cardiovascular conditions, many state health departments and school-based athletic programs have implemented cardiac screening programs as a requirement for competitive athletic eligibility in schools. Screenings include an electrocardiogram (ECG), which can detect ARVC, LQTS, SQT syndrome, and some cardiomyopathies (Caselli, Attenhofer Jost, Rolf, & Pelliccia, 2015). Recent literature has debated whether or not pre-participation screenings are over-diagnosing individuals, but others have argued that this type of screening of young athletes has allowed detection of an increasing number of true diagnoses before a cardiac event has occurred in an individual (Maron et

al., 2004). If a positive screen is further diagnosed as a syndrome at risk for SCD as a common possible outcome, medical management will be enforced (Maron et al., 2016b). In many cases, guidelines state that these individuals should be disqualified from competitive sports and should make lifestyle changes in their exercise activities.

The Bethesda Criteria is a set of guidelines and recommendations that have been released in three revisions, all sponsored by the American College of Cardiology (ACC). The most recent document was released in 2005 for the trained athlete with an identified cardiovascular abnormality that significantly increases the risk for SCD. Specifically, the 2005 criteria provide recommendations regarding eligibility for competition in organized sports (Pelliccia et al., 2008). This set of guidelines is intended to be used as a tool for physicians and medical professionals when presenting medical management recommendations to a newly diagnosed patient. The 2005 revision was fueled by advancements in medical management and the SCD awareness in the community and public media.

Within the Bethesda Criteria, pre-participation screening is discussed for college and high school athletes. The report explained the recommendations for testing options such as echocardiography, electrocardiography, and other forms of imaging (Maron et al., 2004). The majority of the guidelines are somewhat specific by basic etiology of cardiac diseases. Their associated recommendations are based on information known in 2005, now more than ten years in the past. For example, the guidelines state that athletes with hypertrophic cardiomyopathy should be excluded from most competitive sports, with the exception of class 1A sports, which include billiards, bowling, and golf. Similarly, athletes with arrhythmia disorders including LQTS and SQT syndrome are to receive

strict recommendations that include restriction of all sports except for 1A category activities. A diagnosis can differ depending on age, genetics, and presence or absence of an ICD or pacemaker (Pelliccia et al., 2008). The Bethesda Guidelines are often thought by some cardiologists to be overly strict and non-specific to the patient.

In 2015, the American College of Cardiology (ACC) and American Heart Association (AHA) released a scientific statement regarding eligibility and disqualification recommendations for competitive athletes with cardiovascular abnormalities (Maron et al., 2015). The main intention of these organizations was to present a readily usable set of recommendations and guidelines addressing criteria for eligibility for and disqualification from organized competitive sports due to high risk cardiovascular conditions in hopes of promoting the health and safety of young athletes by preventing sudden death in the young. As an update to the 36th Bethesda Conference, the present document serves to adapt to new technology, motivations, and awareness for athletes at risk for SCD (Maron et al., 2015).

The 2015 guidelines classify sports based on their static and dynamic components. See Appendix D for ACC classification of sports activities. The static component is related to level of endurance, or the percentage of a maximal voluntary contraction of any muscle in the body. Sports with a higher static component include intense sports such as rock climbing and water-skiing, each requiring longer muscle contractions. The dynamic component includes how regularly the large muscle groups of the body are being used. More dynamic sports examples include boxing and rowing. In the document, sports are also classified by their intensity to increase the risk of intracranial hemorrhage and/or loss

of consciousness, which is important for individuals undergoing treatment with an anticoagulant agent (Maron et al., 2015).

The second section addressed pre-participation screening in competitive athletes. Screening is recommended to include personal medical history and family history, plus a physical exam. Any suspicious findings may result in further testing, including a stress test, ECG, or echocardiogram. The authors noted that universal ECG testing might be considered in small high school and college populations. Large-scale general cardiac screening with ECGs at universities and public events is not yet recommended (Maron et al., 2015).

The 2015 updated guidelines specified recommendations by type of condition, including cardiomyopathies, congenital heart disease, valvular heart disease, hypertension, aortic diseases, coronary artery disease, channelopathies and arrhythmogenic conditions. The cardiomyopathy task force focused on HCM and ARVC, and recommended that those individuals with symptomatic HCM should not participate in most competitive sports, with the exception of low intensity activities (Class IA sports). Genotype-positive phenotype-negative HCM patients could participate in more competitive sports (Class IIa), especially in the absence of HCM-related sudden death in the family history. All patients with HCM were encouraged to consider pharmacological agents such as beta-blockers to control cardiac-related symptoms (Maron et al., 2015).

Much stricter guidelines were recommended for ARVC, with the 2015 recommendations stating that anyone with a definite or even possible diagnosis of ARVC should not participate in most competitive sports. Low intensity sports (Class IA) are an exception. In addition, ICDs should not be placed in athletes with ARVC or HCM for the

sole purpose of participating in high-intensity sports because of device-related complications (Maron et al., 2015).

The 2015 recommendations combined all channelopathies into one set of recommendations. For symptomatic athletes with any suspected or diagnosed channelopathy condition, the ACC/AHA experts recommended that athletes be restricted from all competitive sports until a comprehensive evaluation has been completed. Asymptomatic athletes, or those who are genotype-positive and phenotype-negative, could be allowed to participate in all competitive sports. This is only if precautionary measures are taken into consideration, including avoidance of QT prolonging drugs in patients. Electrolyte and rehydration replenishment was recommended as critical medical management, and the implementation of an emergency action plan, including access to an automated external defibrillator (AED), was included as precautionary measures that should be taken (Maron et al., 2015).

For channelopathies, the updated guidelines state that previously symptomatic patients who have been asymptomatic while on treatment for at least three months with either medication or an ICD may be eligible for competitive sport participation. This is true for all Class IIb sports except for swimming (Maron et al., 2015). Patients should be closely monitored and followed up by their medical staff throughout participation.

The final portion of the updated guidelines included recommendations regarding drugs and performance-enhancing substances, emergency action plans, SCD triggered by innocent blows to the precordium, and legal aspects of medical eligibility and disqualification (Maron et al., 2015). When considering the entire set of guidelines, nowhere in the document is there a task force directive on how these recommendations

should be communicated to patients. Similarly, the recommendations do not state which medical professionals are responsible for communicating these guidelines to patients (Pelliccia et al., 2008).

The 2015 ACC/AHA recommendations only apply to competitive athletes in competitive sports (Maron et al., 2016a). No defined guidelines for diagnosed patients who are non-competitively active have been published to the best of our knowledge, which is troublesome since the athlete's heart is under notably different stress than those active individuals without a high physical demand during their exercise activities. This difference may necessitate a new set of guidelines for the typical individual who wishes to maintain a healthy exercise regimen after a diagnosis of a genetic cardiovascular condition that includes susceptibility to SCD.

1.4 Impact of a Diagnosis on Psychosocial and Activity Lifestyle Changes

Previous research has shown that a diagnosis with a serious cardiac disease can lead to sensitive psychosocial effects in patients (Rosman et al., 2014). A study performed on adolescents with heart disease showed that individuals present with a variety of self-perceptions regarding behavioral and health-related quality of life (Mussatto et al., 2014). Another study looked more closely at inherited cardiac conditions and psychosocial implications of pre-symptomatic genetic testing (Ormondroyd et al., 2015). This study provided rich data that supported the perceptions of a positive result, and the difficulties of requiring major lifestyle changes. In addition, these authors determined that families might require more psychosocial support or counseling about genetic test information and medical management recommendations in order to alleviate some emotional challenges and physical limitations that they may face.

Despite the evolution of the Bethesda Criteria in 2015 and support groups for SCD, research studies have shown significant psychosocial impacts on athletes after they have received a potentially lethal cardiac disease diagnosis (Asif et al., 2015a). These authors reported patients' feelings of avoidance and hyper-arousal about their conditions. The highest risk individuals for significantly negative psychosocial impact included those who were permanently disqualified athletes. Those who were able to participate in certain levels of activity had less emotional impact, suggesting the potential effects of the strict Bethesda guidelines on patients who are not competitive athletes and who may have a less serious diagnosis, which still limits their exercise practice. The authors concluded that these disqualified athletes tended to avoid conversations regarding their diagnosis and may require further emotional and informational support than is currently being provided to them.

Another similar study looked at the role of medical professionals in an athletic patient's diagnosis of a potentially lethal cardiac disease. The risk for psychosocial distress was increased for those with higher level of competition, those with complete restriction from activity, and athletes with unanticipated outcomes or failed procedures in their medical management. An important conclusion from this study was that medical professionals typically lacked emotional support counseling skills and clarity in their information regarding procedures and medical management (Asif et al., 2015b). Minimal research exists that studies the psychosocial effects of a lethal cardiac disease diagnosis on non-competitively exercising adults.

The implantable cardioverter defibrillator (ICD) offers protection against SCD by terminating potentially lethal arrhythmias (Maron & Maron, 2016c). ICDs have been

used for both primary and secondary prevention in patients with cardiomyopathies, arrhythmia conditions, and previous history of unexplained syncope. Primary prevention is defined as ICDs for those who are at risk for a first arrhythmia. Secondary prevention applies to those who have previously experienced an arrhythmia or have experienced cardiac arrest without an ICD at that time (Carroll & Arthur, 2010). Risks associated with ICDs can include inappropriate shocking when unnecessary as well as delayed shock during cardiac arrest. Despite their limitations and risks, ICDs have allowed patients who would have been previously limited from all activities to take part in moderate exercise practices (Gaba et al., 2016).

Carroll and Arthur (2010) studied the psychosocial effects of patients receiving their first ICD for both primary and secondary prevention of SCD. The researchers measured outcomes of levels of anxiety, uncertainty, and optimism before and after the implant. Individuals experienced significant uncertainty before receiving the ICD. Optimistic disposition and normal anxiety were common themes also identified before the ICD was implanted. However, anxiety levels measured higher in secondary prevention patients than in primary patients who had not yet experienced a cardiovascular episode due to their condition. Overall, these authors described an emphasis on the psychosocial and informational needs of these patients with new ICDs. They concluded that patients with ICDs should be adequately educated on the risks, expectations, and functions of the device, especially prior to exercise or activity.

A consistent theme across many of these publications that focus on athletes was that competitive individuals experience the most negative psychosocial impact when they are disqualified from their desired sport. The fact that their biggest stress-relieving

activity, passion, or even career is taken away from them is heartbreaking for most. Research has shown that some athletes choose to continue participating in sports both with and without ICDs. These patients mostly showed low rates of cardiac events and no deaths. Minimal research has been published on how similar exercise restrictions on individuals who are not competitive athletes can emotionally and physically affect them. Adults who have a typical exercise practice but do not participate competitively may experience different psychological effects after a diagnosis (Johnson & Ackerman, 2012). A potential lethal cardiac disease diagnosis that is associated with SCD is psychologically traumatic for any patient, and requires adequate counseling and guidance to provide the safest and most adequate medical management and support for each individual patient.

1.5 The Effectiveness of Current Guidelines and Exercise Recommendations

As previously stated, the exercise recommendations for athletes remain strict, conservative, and non-personalized. In most cases, adherence to the Bethesda Criteria results in disqualification from all competitive sports and activities. To our knowledge, no current guidelines exist for average active adults at risk for SCD who exercise at or below the suggested 150 minutes each week as recommended by the AHA to benefit one's health and well-being (Cheung, Laksman, Mellor, Sanatani, & Krahn, 2016). Many patients at risk for SCD find it critical to balance the benefit of physical activity in cardiovascular disease with their risk for SCD. Previous research by Canadian healthcare providers have reported the recommendations that they share with patients (Roston, De Souza, Sandor, Sanatani, & Potts, 2013). These findings indicated that the majority of cardiac care providers who participated in the study only partially communicated current exercise recommendations. Physical activity recommendations for many of the congenital

physical and electrophysiological heart defects varied considerably. This valuable study supports the hypothesis that the medical professionals are practicing inconsistent communication of essential medical management information to their patients. Thus, further research is needed on patient experience and understanding of their exercise recommendations and medical management safety.

Research in genetic causes of these cardiovascular conditions has improved tremendously, and previously generalized diagnoses are now categorized by the specific mutation within a given gene or the genetic etiology of each individual case. Because of this, each patient should have his/her own personalized medical management recommendations based upon the physical makeup which includes the specific genetic etiology of disease, age, weight, ethnicity, environment, and other relevant factors such as family history.

A recent research study of the 2005 Bethesda guideline limitations in exercise restrictions for patients with inherited cardiac conditions stressed the impracticality and ineffectiveness of such rigid guidelines. With such heterogeneous syndromes, individualized patient-centered appraisal and care was recommended for the future. New general guidelines through the 2015 AHA and ACC stressed the importance for movement towards personalized medicine in cardiology (Hammond-Haley, Patel, Providencia, & Lambiase 2016).

The decision to continue or discontinue an exercise practice is ultimately up to the patient or their family in the pediatric age group. Their decision is influenced by factors including past experiences, personality type, family or peer influence, love for activity, and understanding of medical recommendations. A research study of athletes with LQTS

showed that 37% of patients chose to remain in competitive athletics. Some of these patients had ICDs, and it is important to note that none of these athletes had a sport-related cardiac event. A conclusion from this study supported that current guideline recommendations for disqualification may be excessive for disease (Johnson & Ackerman, 2012). Since this study focused on competitive athletes, it may be true that even these guidelines may also be excessive for non-competitive active adults who experience much less physical stress than athletes.

Conclusions from these studies reflect that current recommendations apparently are not adequately or consistently communicated, enforced, or followed by at-risk individuals. The field of cardiovascular SCD conditions needs updated guidelines that focus more individually on exercise recommendations appropriate for each specific condition, and could benefit from a personalized medical management interpretation that is unique to each individual case. Older guidelines that seem overly rigid and strict may be too excessive, and could lead those at risk for SCD to take part in activities that could lead to cardiac arrest. The current state of knowledge in 2017 should bring together the multiple factors that influence exercise management for these diagnoses, including gene-specific causes and genotype-phenotype correlations; cardiac research; technologies such as ICDs and pacemakers; and the availability and knowledge of cardiovascular genetic counselors to update guidelines that could then be effectively communicated to all patients at risk for SCD.

1.6 Role of a Genetic Counselor in Cardiovascular Genetics

The role of a genetic counselor has been clearly described. The Genetic Alliance (Genetic Alliance, 2009) has identified genetic counselors as a central resource of

information about genetic disorders for patients, other healthcare professionals, and the general public. In general, a genetic counseling session aims to increase the patient's and family's understanding of a genetic condition, including management and testing options. Genetic counselors are trained to identify the psychosocial triggers of a person diagnosed with a genetic condition to help those patients cope with potential implications such as future possible outcomes, and to reduce overall anxiety due to the diagnosis (Genetic Alliance, 2009).

All genetic counselors are trained to develop exceptional skills in patient education and psychosocial processes. These skills are performed in a variety of concentrations including but not limited to pediatrics, cancer, cardiovascular, prenatal, and neurology specialty clinics. Genetic counselors are expected to educate patients on the benefits, risks, limitations, and potential outcomes of genetic testing; interpretation of results and their implications; medical management recommendations; and outside resources for supporting the patient and family. They also have unique skills to interpret and manage the psychosocial elements that may arise in a session when a positive genetic test result is revealed (Uhlmann, Schuette, & Yashar, 2011).

Cardiovascular genetics is particularly interesting for genetic counselors. Genetic testing is typically aimed at reducing uncertainty, but cardiovascular genetics can leave patients feeling uncertain about their risks for cardiac events, medical management, and risk for family members if those implications are not fully addressed. Patients have reported experiencing psychological distress in regards to not only the diagnosis, but also how to manage that diagnosis (Ackerman et al., 2011). Various research studies have shown the significant elevated distress and negative psychosocial impact of a positive

genetic test for a condition that increases risk for SCD (Mussatto et al., 2014; Asif et al, 2015).

Other genetic counseling concentrations, such as cancer genetic counseling, have clear testing and management guidelines that counselors use when educating patients about a positive test result. These include the NCCN guidelines for management of an individual with a positive BRCA1 or BRCA2 test result, or of an individual who has been genetically diagnosed with Lynch syndrome as a hereditary type of colon cancer, as well as similar guidelines from professional organization such as The American Society of Clinical Oncology (ASCO) and the American College of Obstetrics & Gynecology (ACOG). Similar management guidelines exist for cardiovascular genetic testing through the American Heart Association and Heart Rhythm Society; however, no definitive structure exists for cardiovascular genetic counselors to discuss topics such as exercise management which today are beyond the topics of genetic testing and implications of genetic test results for the patient and the patient's family (Ackerman et al., 2011). All genetic counselors have the skills and expertise to address the psychosocial implications that come with cardiovascular genetic counseling, including the patient information and implications about exercise recommendations if those recommendations exist for the specific SCD condition.

This study addresses the needs of non-competitive active adults who have been diagnosed genetically with an SCD condition that may call for restriction of their exercise activities. We proposed that most of these patients would be less than satisfied with the exercise recommendations that they were given, as no clear guidelines yet exist for reconciling a gene-specific SCD condition with personalized exercise recommendations

for this group of active adults. The study could result in a call for exercise guidelines as medical management guidance to be formally included within the domain of cardiovascular genetic counselors, which could benefit this class of patients with genetic conditions.

Chapter 2

Exercise Recommendations for Active Adults at Risk for Sudden Cardiac Death: “Can I Continue to Exercise?”¹

Section 2.1 Abstract

Sudden arrhythmic death syndrome (SADS), is due to genetic conditions associated with cardiac ion channelopathies and cardiomyopathies. Many of these conditions have exercise guidelines regarding eligibility and disqualification recommendations for competitive athletes through the American Heart Association and the American College for Cardiology. This study investigates how medical professionals communicate these exercise recommendations to patients, and in turn, how well patients understand these recommendations. The study also explores motivations for either continuing or discontinuing an exercise practice after a diagnosis. A questionnaire was distributed through cardiac disease-focused support groups and organizations. Data from 67 individuals with a SADS condition were analyzed for possible associations and significant differences. Results demonstrated that intensity of exercise on a 1 to 9 scale decreased from 4.9 to 3.3 after a diagnosis ($p < .01$). Time spent exercising decreased from 204 minutes per week to 142 min/week after a diagnosis ($p < .01$). Patients were significantly more likely to decrease activity intensity when they felt confident with the communication of their exercise recommendations and how well they understood them.

¹Baker, K., Walker, P., Asif, I., & Hill-Chapman, C. Manuscript to be submitted to *Circulation*, 2017.

This study showed that individuals felt poorly informed regarding their exercise recommendations and often take on heavy emotional burdens due to restrictive guidelines. The respondents widely expressed that quality of life is an important factor in their decisions about exercise, and that continuing exercise is a major component of a healthy lifestyle despite risks associated with SADS. Involving genetic counselors in the discussion of exercise guidelines is recommended to address the psychosocial outcomes of a recent diagnosis. This addition to the management of individuals with a SADS diagnosis is likely to increase awareness of how exercise recommendations are managed and how well patients understand that their personalized exercise plan will promote a safer healthcare management plan for each patient.

Section 2.2 Introduction

Sudden arrhythmic death syndromes (SADS) are genetic heart conditions that can cause sudden cardiac death (SCD) in individuals where a structural heart defect may or may not be present. Some of the genetic cardiovascular diseases include but are not limited to cardiomyopathies and channelopathies (Sheikh et al., 2015). Cardiomyopathies refer to diseases of the heart muscle, where muscle tissue becomes thickened and rigid, making the heart weaker and prone to arrhythmias and even SCD. Channelopathies are diseases that result from the dysfunction of ion channels located in the membranes of cells, and specifically in cardiac tissue. Channelopathies can lead to irregular heart rhythms, syncope, arrhythmias and SCD. LongQT and Brugada syndrome are two of the more common ion channelopathies associated with a risk of SCD (Napolitano et al., 2012).

Genetic testing for these heart conditions associated with SADS has improved dramatically, and multigene panel testing is available for symptomatic patients or those with a family history. Since many of these conditions may show no clinical signs on echocardiogram (ECG) or echocardiography, genetic testing has become the gold standard for diagnosing these conditions. Patients at risk are encouraged to seek genetic counseling for analysis of family history and pre-test counseling when indicated (Ormondroyd et al., 2014). Since both cardiomyopathies and channelopathies display reduced penetrance and variable expressivity, not everyone who carries a mutation will develop symptoms (Cirno & Ho, 2014). In addition to a genetic and clinical diagnosis, the patient's age, sex, and lifestyle choices influence the risk for developing symptoms (Ackerman et al., 2011).

Despite health benefits that typically result from exercise in most individuals, the subset of individuals who are at risk for SCD must weigh the health risks that come with exercise. Research shows that individuals at risk for SADS may have a higher risk for an adverse cardiac event during exercise (Maron et al., 2016b). In 2015, the American College of Cardiology (ACC) and American Heart Association (AHA) released a scientific statement regarding eligibility and disqualification recommendations for competitive athletes with cardiovascular abnormalities. This statement presented a readily usable set of recommendations addressing criteria for participation or disqualification from organized competitive sports due to high-risk cardiovascular conditions in hopes of promoting the health and safety of young athletes by preventing SCD (Maron, et al., 2015).

The 2015 statement classifies sports based on their static and dynamic components, and includes activity recommendations and medical management guidelines specific to each cardiac condition. See Appendix D for ACC sport classification. This document targets a small portion of the entire population at risk for SADS, and there are currently no guidelines that are specific to the larger population of active adults who are non-competitive athletes.

Previous research has shown that a diagnosis with a serious cardiac disease can lead to sensitive psychosocial effects in patients (Rosman et al., 2014). Presently more commonly used in athletes is the ICD, which is a piece of technology that offers protection against SCD by terminating potentially lethal arrhythmias (Maron & Maron, 2016c). The ICD can benefit psychosocial impact by making exercise guidelines less strict, but can also increase patient anxiety due to fear of shock during a cardiac event and even inappropriate shocking (Gaba et al., 2016).

As stated previously, to our knowledge no current formal exercise guidelines exist for average active adults at risk for SADS. However, many patients at risk for SADS find that balancing the benefit of physical activity with their risk for cardiac events is critical to their well-being. Previous research in Canada surveyed Canadian cardiac healthcare providers who indicated that they only partially implement current exercise recommendations (Roston et al., 2013). Another study looked at competitive athletes with LQTS, and found that 37% of those individuals chose to remain in competitive sports despite their diagnosis (Johnson & Ackerman, 2012). These studies have led to conclusions that current exercise recommendations may not be adequately communicated or are not followed by a significant portion of these at-risk individuals.

No currently recommended guidelines exist stating which medical professionals should be sharing exercise recommendations with patients. After a SADS diagnosis, medical management includes not only exercise guidelines, but also likely includes a psychosocial burden. Other information such as potential risks for family members, patient education on the etiology of the disease, and further medical management are critical components to also be addressed at the time of diagnosis. Recognizing that these are all topics that require an ample amount of time with patients, perhaps the medical community should seek additional healthcare team members to assist with the diagnosis and translating the full implications of that diagnosis for the patient and her/his family.

Genetic Counselors are individuals who assess individual or family risk for a variety of inherited conditions including SADS conditions (Genetic Alliance, 2009). Cardiovascular genetic counselors are growing in numbers, with more genetic counselors present in cardiology clinics as their fulltime responsibility and others interacting on a regular basis with cardiologists who refer their patients to specific genetic counselors with cardiovascular training and expertise. Cardiovascular genetic counselors may be appropriate individuals to address SADS medical management, exercise recommendations, and the psychosocial impacts that may follow a diagnosis of a genetic cardiac condition that increases the patient's risk of SCD (Ackerman et al., 2011).

Regardless of which medical professionals are communicating medical management and exercise recommendations for those at risk, increased awareness of how exercise recommendations are being communicated to patients and how well patients understand their medical management should promote a safer and more personalized

healthcare plan for patients, while improving the psychosocial outcomes of a recent diagnosis.

Section 2.3 Materials and Methods

This research study collected quantitative and qualitative data from active adults who have a genetic condition that puts them at risk for SCD. Participants were recruited through condition-specific support group organizations and Facebook groups targeted for families affected by the previously described conditions. Individuals over the age of 18 who have had a diagnosis associated with increased risk of SCD or sudden arrhythmic death syndrome (SADS) and could comprehend the medical and technical information in the questionnaire were eligible to participate in this study.

An invitation to participate (see Appendix A) as well as a link to the survey was hosted on surveymonkey.com, with access to the survey distributed through several venues. The SADS Foundation and Hypertrophic Cardiomyopathy Association were contacted via email to explain the purpose of the study and inquire if they were willing to invite their members to participate. These organizations were willing to invite their members to participate and did so via their respective electronic mailing lists. In addition, a link to the survey (see Appendix B) was posted on various Facebook groups throughout September, 2016, and was available until December 31, 2016.

The questionnaire consisted of multiple choice and open-ended questions that investigated demographic information for each of the participants. It also explored the diagnostic process; pre and post-exercise activity including length of time and type of sport or activity; and number of cardiac events. Other questions queried topics regarding ICDs, medication, and confidence in understanding of guidelines. Open-ended questions

explored the psychosocial affects following a diagnosis and discussion of appropriate post-diagnosis activities.

Quantitative analysis was performed with SPSS software, version 24, using a dependent samples analysis technique to assess trends before and after a diagnosis. For categorical dependent variables, non-parametric analysis techniques were employed, including logistical regression, test of marginal homogeneity, and chi-square test for independence. For continuous quantitative dependent variables, repeated measures analysis of variance (ANOVA) was used to assess for statistical significance, set at $p = .05$ or less. Descriptive statistics were calculated by Excel software for percentages and basic information.

Qualitative questions from the survey were separately coded by two independent individuals (first and second authors) using grounded theory methods. After individual coding, findings were discussed to reach consensus on basic themes that emerged from the qualitative responses. This research study was approved by the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC, in June, 2016.

Section 2.4 Results

Eighty-seven individuals participated in the survey. Sixty-seven individuals met criteria to be included in the study and completed the survey. Respondent demographics are displayed in Table 2.1. Of the 67 respondents, 17 (25%) were male and 50 (75%) were female. The mean age of the participants was 48 years, with a range of 18 to 72 years of age. The majority of respondents were Caucasian ($n = 61$, 91%) and had completed at least some college ($n = 55$, 82%). The majority of participants had been

diagnosed with HCM ($n = 34$, 51%), and 17 individuals had been diagnosed with LQTS (25%), 15 with Brugada syndrome (22%), and one with LVNC (2%). The average age at time of diagnosis was 39 years.

Table 2.1
Demographics

	Frequency (<i>n</i>)	Percentages (%)
Gender		
Male	17	25%
Female	50	75%
Ethnicity		
Caucasian	61	91%
Asian/Pacific Islander	4	6%
African American	0	0%
Hispanic/ Latino	0	0%
Mediterranean	2	3%
Highest Level of Education		
Did not complete High school	3	5%
High school diploma or GED	7	11%
Some college	9	14%
Associate's Degree	8	12%
Bachelor's Degree	20	31%
Some Graduate School	4	6%
Graduate Degree (Masters, PhD, MD, etc.)	14	22%
Type of Diagnosis		
Hypertrophic Cardiomyopathy	34	52%
Long QT Syndrome	17	26%
Brugada Syndrome	15	21%
LVNC	1	1%
Do you have an ICD?		
Yes	30	44%
No	37	55%

Statistical analysis was performed using SPSS software. Statistical significance was set at $p = .05$ or less. We analyzed minutes of activity before and after a diagnosis, and type of activity before and after a diagnosis using paired T-Tests. Activity level was coded on a ranking scale of one unit to nine units (one being least intense level of activity, and nine being most intense) using the static/dynamic activity chart from the AHA guidelines (Maron et al, 2015). The mean activity intensity before a diagnosis was 4.90 units ($SD = 2.41$), and after a diagnosis was 3.32 units ($SD = 2.44$) ($p < .01$). The mean minutes of exercise before a diagnosis was 204 minutes per week ($SD = 178.1$), and was 142 minutes per week ($SD = 127.5$) after a diagnosis ($p < .01$).

Repeated measures ANOVA test compared the difference in activity intensity before and after a diagnosis and the difference in minutes of exercise before and after a diagnosis against the following four variables:

- the confidence with participants' understanding of exercise recommendations (Likert Scale 1-5). See Figure 2.1;
- satisfaction with the medical professional's communication of exercise recommendations (Likert Scale 1-5). See Figure 2.2;
- presence or absence of ICD; and
- educational level by category.

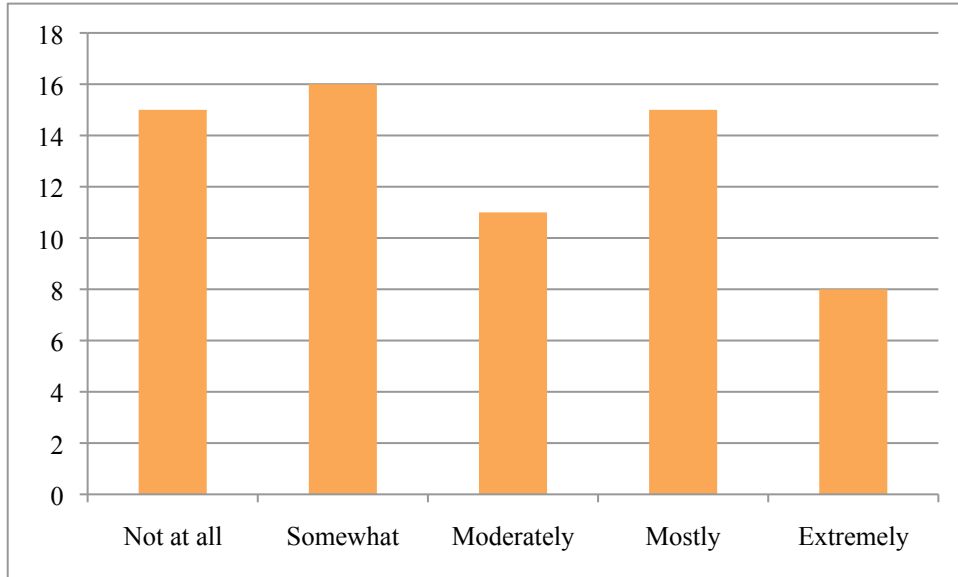


Figure 2.1. Confidence in Understanding Exercise Recommendations (n = 65)

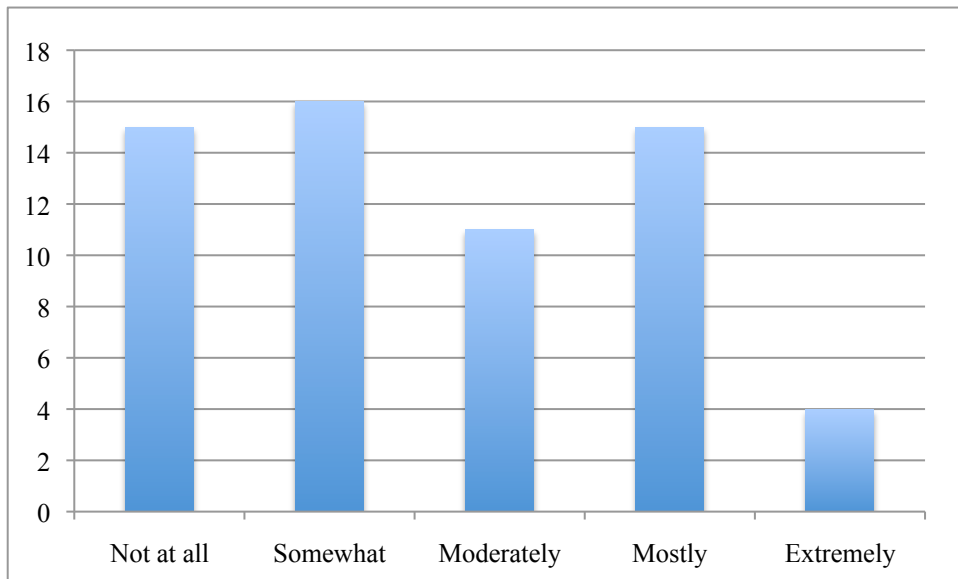


Figure 2.2. Satisfaction with Communications about Exercise Recommendations. (n = 61)

Table 2.2 displays the statistical outcomes of eight comparisons, three of which were found to be statistically significant. Individuals who were more confident with their

understanding of exercise recommendations were statistically significantly more likely to decrease activity intensity after a diagnosis ($p < .01$). Individuals who were more satisfied with the communication of exercise recommendations by medical professionals were statistically significantly more likely to decrease exercise intensity after a diagnosis ($p < .01$). Those with a higher level of education exercised for fewer minutes after a diagnosis also showed statistical significance ($p < .01$). All other repeated measures ANOVA comparison tests did not suggest statistical significance. See Appendix C for the full ANOVA comparison statistics.

Table 2.2 ANOVA Comparisons		
	Change in Exercise Intensity	Change in Exercise Time (min)
Confidence with Understanding of Recommendations	$p = .037^{**}$	$p = .160$
Satisfaction with communication of recommendations	$p = .042^{**}$	$p = .205$
Presence of ICD	$p = .793$	$p = .075$
Educational Level	$p = .770$	$p = .039^{**}$

** = statistically significant

Analysis of qualitative data showed three major themes when evaluated independently by two authors. These main themes were identified as follows:

- Individuals feel poorly informed regarding their exercise recommendations;
- Participants take on differing levels of emotional burden with exercise recommendations; and
- Exercise is a key component to quality of life in these active individuals.

Individuals feel poorly informed regarding their exercise recommendations.

Some individuals were told to simply listen to their body when exercising and to be aware of signs and symptoms when they appear. Many respondents felt conflicted on what is considered enough exercise for a healthy lifestyle, and what is considered too much that puts them at a higher risk for cardiac event.

Now that my 18 year old body is gone, I do feel the need to exercise, but it is confusing. What is the max I can push myself to? I don't know, and I don't think science knows either. I find that walking, and yoga are my safest bets. Yoga has been strongly recommended by my Electrophysiologist not only because he feels it's a safe level of activity, but for my emotional/mental well being as well.

– 26yo female with LQTS

However, most respondents stated that they were never given exercise recommendations and had to seek out advice on their own. “I had to ask for advice but nothing was offered. Basically don't lift weights and no running.” - *37yo female with HCM*. Figure 2.3 shows which resources patients utilized after a diagnosis to seek out exercise recommendations.

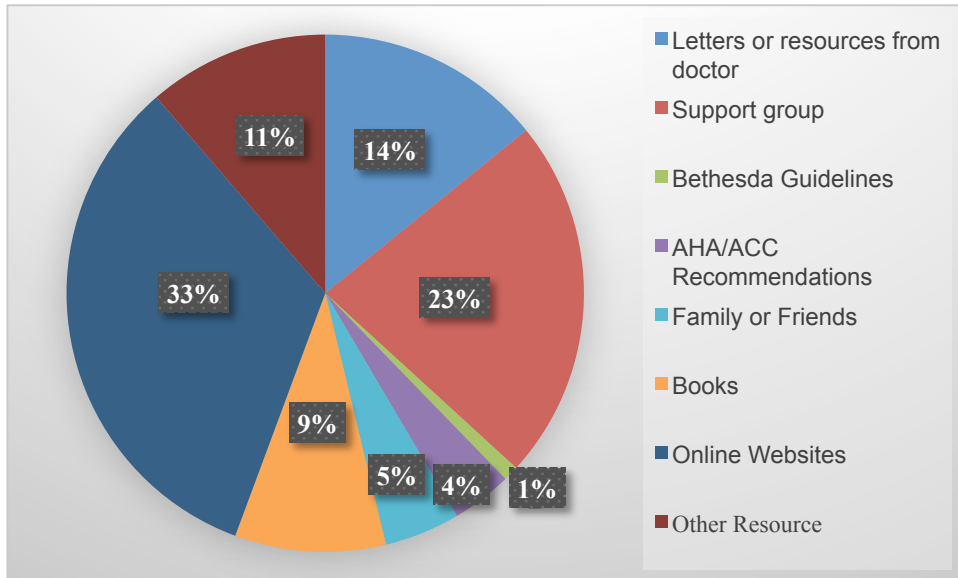


Figure 2.3. Resources Used by Patients to Better Understand their Exercise Recommendations

*Other includes: "No resources used," "Doctor," & "Personal common sense"

A large segment of participants stated feeling confused, conflicted, and that they did not understand guidelines.

Patients feel differing levels of emotional burden with exercise recommendations.

Many participants claimed to be devastated with their new diagnosis leading to different exercise recommendations, while some others stated that they felt reassured with their diagnosis with no comment about exercise recommendation. For some, exercise was the main source of stress relief, and participants stated feeling shocked and surprised with the information. Many also felt frustrated with the strict limitations and overwhelmed with the recommendations that are not directed for non-competitive athletes.

“I was sad because I'm a pretty active person and also feeling that I wasn't fully informed about other activities that I could do like hiking, climbing, yoga, weight lifting and etc.”

- 27yo female with LQTS

Participants admitted to living in fear of having a cardiac event during exercise.

That fear and the potential risk of leaving family members behind is an emotional burden that comes for some with exercise recommendations.

“Anxious, easy to make excuses not to exercise. Very hard to stay motivated, unsure of what safe boundaries are for me personally. Want to fight against it as want to be healthy but also want to be here for my three kids so not willing to risk much.”

- 48yo female with LQTS

Exercise is a key component to quality of life in these active individuals.

Multiple participants responded with willingness to accept risks of exercising, as they wish to live life on their own terms by participating in activities that they love the most. Many participants stated the importance of staying in shape and exercising to keep their weight down to improve their overall health and well-being.

“I love exercise. It's always been a part of my life. I want to be fit and healthy. I feel good when I exercise. I just don't push myself as hard as I used to. It's a challenge finding the right balance.”

- 37yo female with Brugada syndrome

In addition, many feel that exercise is a stress reducer that works to better mental health.

By removing a stress reliever at a time where emotional burden is high, many individuals feel even more emotionally impacted.

“My motivations for carefully resuming activity now are psychological.

Exercise helps me lowering my anxiety, it improves my tolerance to stress and helps me trust my body and my meds again. I still stick to medical

advice and I don't engage in competitive sports but I've realised I need to stay active if I want to feel healthy.” – 39 yo female with LQTS

Section 2.5 Discussion

From these results, we can conclude that both the intensity of activity and the time spent exercising changed significantly after a diagnosis of a condition that puts a patient at risk for SCD. However, our analysis shows a statistically significantly greater difference in the intensity of activity after a diagnosis than in the time spent exercising. In other words, participants were more likely to change their activity type than to change their overall time spent exercising. We observed that those who were more confident with their understanding of exercise were more likely to decrease their exercise intensity after a diagnosis. Similarly, those who felt more satisfied with the communication of exercise recommendations by medical professionals also were more likely to decrease their exercise intensity after a diagnosis.

This important finding identifies key factors, which can motivate a patient to change an exercise practice after a diagnosis. We measured that understanding and communication of guidelines do not impact the time spent exercising. However, both communication of and understanding of exercise recommendations can have a significant impact on patient's decision to decrease their exercise activity intensity. Thus, if patients better understand guidelines that are communicated to them, they are more likely to exercise at safer intensity levels or to choose entirely new exercise activities after a diagnosis. These findings support the implication that improved patient care may be possible through key communications about exercise alternatives and could lead to perhaps more responsible medical management by the patient.

We found that individuals with higher educational levels exercised for a shorter time period after a diagnosis than they had before the diagnosis. This interesting finding could potentially be explained by their higher education level and a better understanding of exercise guidelines. This finding also highlights the importance of patient education in the clinic, and the need to share the information with every patient in a way that she/he can comprehend the information and take appropriate action to adapt their new exercise regimen to their new diagnostic status. Discussions tailored to the patients' education levels could prevent those with less education from putting themselves at a greater risk for cardiac events or SCD. Medical professionals must understand the patient's ability to understand the implications of a diagnosis and then adequately educate each patient to ensure their understanding of appropriate exercise recommendations. Improving patient understanding and building patient confidence with medical guidelines when they leave the medical offices should help each patient correctly understand and utilize the information that they have learned.

A recent diagnosis with exercise recommendations has the potential to have serious psychological impact on any patient. Extensive previous research has been performed with competitive athletes (Asif et al., 2015a; Asif et al., 2015b; Maron, B. et al, 2016a) but minimal research, if any, has been published and made available as clear guidelines for non-competitive active adults who simply wish to live an active and healthy life. Our study demonstrated similar findings for non-competitive individuals, and that most of them feel devastated, confused, depressed, sad, scared, and/or conflicted after a diagnosis that requires new limitations on their exercise program but with no formal guidelines. We found that non-competitive active adults felt emotionally burdened, similar to the feelings

of competitive athletes in similar circumstances. Medical professionals should be aware of this emotional burden when providing medical management guidelines, as patients do not have to be exercising competitively to have a severe psychological impact.

Our results agreed with previous research regarding the emotional burden that comes with a diagnosis, which requires exercise guidelines and limitations (Asif et al., 2015a; Asif et al., 2015b). Most importantly, individuals stated strong feelings of confusion, insecurity, and confliction in regards to the new exercise recommendations. This is the first research study to our knowledge that focuses on the communication and understanding of exercise recommendations for non-competitive active adults with a diagnosis that puts them at risk for SCD or SADS, and how this information impacts their decision to continue or discontinue an exercise practice. Our findings demonstrate that additional research would likely benefit this medical field to ensure that patients and their physicians are understanding appropriate exercise recommendations. Medical professionals should adequately educate their patients about their exercise guidelines. This can be done by ensuring an ample amount of time is available, by using teaching skills at various educational levels, and by paying attention to the psychosocial burden felt by the patient to provide more thorough care and better resources for patients.

Genetic counselors are beginning to enter the field of cardiology. They spend, on average, about 45 to 60 minutes with each patient to complete risk assessments, to gather a comprehensive three-generation family history, and to educate the patient on genetic testing and medical management (Uhlmann et al., 2011). Our study suggests that genetic counselors could be appropriate medical professionals to share some of the medical management responsibilities for these patients, as typically performed in sessions with

patients who have been found to have a genetic colon cancer diagnosis, where management guidelines are published by several professional organizations (NCCN, 2016; Daly et al., 2016). If exercise guidelines are given to the patient from a doctor, genetic counselors can spend more time educating the patient about these guidelines and addressing the patient's psychosocial needs over the course of the counseling session.

Regardless of who begins to take on a deeper role in educating these patients on their medical management, the medical field must become aware of the needs of this group of non-competitive athletes who are at an increased risk for SCD. Improvements have been made on exercise guidelines, and now is the time that critical improvements be made on the forms of communications in the office to ensure the best medical care for our patients when they make decisions about exercise outside of the physician's office. A satisfying sense of understanding their new recommendations for exercise could help to alleviate some of the psychosocial impact that comes with a life-threatening diagnosis, as patients will better understand how to live a safe and active life when they leave the doctor's office.

This study targeted participants who are active members in various support groups. The source of these participants may have created a bias in the results, as patients may have access to more resources and support from these organizations than others who did not respond because they are not connected with any of these support groups. This access to support group information may influence or even bias participants' understanding of exercise recommendations and their decisions to continue or discontinue an exercise practice. The final population demographics were also not representative of the general population. The majority of participants were Caucasians

who had at least some college, obviously a highly educated population who are proactive in seeking out additional resources for themselves. Based on the demographics results, this study did not capture other socioeconomic populations who may be less educated, have poor access to resources, and may be generally less knowledgeable about their medical management recommendations.

Future research should explore various medical professionals' perspectives on communication of exercise guidelines to this class of patients. By interviewing physicians or genetic counselors, the medical community could get a more accurate and comprehensive view of what these professionals are already discussing in their practice, how confident they are in providing patients with exercise recommendations, and how they think this area can be improved. Another area of future research may be understanding the role of an ICD on psychosocial impact and exercise practices after a SADS diagnosis. Our study suggested results trending towards statistical significance based on the impact of an ICD on exercise intensity after a diagnosis, but the number of respondents with an ICD was small, making this is an area to be studied further.

Section 2.6 Conclusions

Our study supports a primary conclusion that individuals with a SADS diagnosis at risk for SCD generally do not feel confident with their exercise recommendations. A lack of patient understanding about exercise recommendations, patient education level, and poor communication of guidelines by medical professionals can affect the duration and intensity at which individuals exercise after a diagnosis. This may impact the safety of the patient when the responsibility is left up to that individual. Since exercise is a key component to quality of life in active adults, exercise recommendations cause

psychosocial burdens on patients after a diagnosis. No clear personalized guidelines exist that specify which medical professionals communicate these guidelines and to what detail they are described. Genetic counselors are medical professionals with a strong medical genetics background and have the psychosocial skills to manage a longer informational session with patients to address this medical management discussion. Improvements need to be made regarding how exercise guidelines are communicated to all individuals at risk for SADS to ensure sufficient patient understanding, as this is an area in which patients feel that they lack information and direction for their post-diagnostic exercise program.

Chapter 3: Conclusions

Our study supports a primary conclusion that individuals with a SADS diagnosis at risk for SCD generally do not feel confident with their exercise recommendations. A lack of patient understanding about exercise recommendations, patient education level, and poor communication of guidelines by medical professionals can affect the duration and intensity at which individuals exercise after a diagnosis. This may impact the safety of the patient when the responsibility is left up to that individual. Since exercise is a key component to quality of life in active adults, exercise recommendations cause psychosocial burdens on patients after a diagnosis. No clear personalized guidelines exist that specify which medical professionals communicate these guidelines and to what detail they are described. Genetic counselors are medical professionals with a strong medical genetics background and have the psychosocial skills to manage a longer informational session with patients to address this medical management discussion. Improvements need to be made regarding how exercise guidelines are communicated to all individuals at risk for SADS to ensure sufficient patient understanding, as this is an area in which patients feel that they lack information and direction for their post-diagnostic exercise program.

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Appendix A: Invitation Letter to Participate in Survey

This study is being conducted as part of the requirements for a Master's of Science degree in Genetic Counseling at the University of South Carolina. We are interested in better understanding your information, thoughts, and opinions about exercising before and after your diagnosis of a heart condition.

You are being asked to participate in this study because you have been previously diagnosed with a cardiac disease that puts you at risk for sudden cardiac death (SCD). The purpose of this study is to explore the communication and understanding of exercise recommendations, and the motivations for either continuing or discontinuing an exercise practice after your diagnosis.

Your participation in this study will increase our understanding on how effectively medical professionals communicate exercise recommendations to their patients at risk for SCD. The results from this study will give insight on how well patients understand their exercise recommendations, while also help us to determine the effectiveness and usefulness of existing exercise guidelines. We believe that the results of this study can – help us better understand patient motivations about why people with your diagnosis choose to continue or to discontinue exercising after their diagnosis so that doctors and other medical professionals can better address the individuals needs of their patients.

Your participation in the study is voluntary. You can skip a question or withdraw from the study at any time. Participating in the study involves completing a short online survey about your experiences before and after your diagnosis. The survey is anonymous, meaning that we will not collect any personal information that could identify you or connect you to your responses. The survey should take about 10 to 15 minutes to complete. Questions in the survey will ask you about your medical and exercise history, how well you understand specific topics. There are also some questions that are open for you to answer with your opinions or in any way that you think is appropriate about your personal experiences and opinions during your diagnosis.

The study is being conducted by Kacie Baker, a genetic counseling student at the University of South Carolina School of Medicine for a Master's thesis project. Peggy Walker, a genetic counselor at the University of South Carolina, is the faculty thesis advisor for this study. If you have any questions about this study, you may contact us:

Kacie Baker, BS
Phone: (908) 278-1203

Email: Kacie.Baker@uscmed.sc.edu

Peggy Walker, MS, CGC

Phone: 803-549-6259

Email: mpwalker11@earthlink.net

For questions about your rights as a participant, you may contact the Office of Research Compliance at the University of South Carolina at (803)-777-7095.

You can access the online survey by clicking the “Next” button below, indicating your consent to participate in this study.

Thank you for sharing your information and insights.

Appendix B: Online Survey

1. What is your gender?

2. What is your age?

3. What is your ethnicity/ancestry? (Please select all that apply)

- Caucasian
- Asian/ Pacific Islander
- African American
- Hispanic/ Latino
- Mediterranean
- Other (please specify)

4. What is your highest level of education?

5. Type of Diagnosis?

- Hypertrophic Cardiomyopathy
- Long QT Syndrome
- Brugada Syndrome
- Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)
- Left Ventricular Non-Compaction Cardiomyopathy (LVNC)

Other (please specify)

6. Age at Diagnosis?

7. Which Medical professionals have you been seen by? (Check all that apply)

- Cardiologist
- Geneticist
- Sports medicine doctor
- General physician
- Genetic counselor

8. I was diagnosed by: (Screening, symptoms, family history, ECG, etc)

- Screening
- Symptoms
- Family History
- ECG
- Other (please specify)

9. Who has discussed exercise recommendations with you after your diagnosis? (Check all that apply)

- Cardiologist
- Geneticist
- Sports medicine doctor
- General physician
- No one has discussed exercise recommendations
- Other (please specify)

10. At what age did you receive exercise recommendations?

11. If you are following current exercise recommendations either partially or completely, at what age did you begin following them?

12. How satisfied were you with the communication of exercise recommendations by medical professionals

- Not at all Somewhat Moderately Mostly Extremely

13. How confident did you feel with your understanding of exercise recommendations?

- Not at all Somewhat Moderately Mostly Extremely

14. Did your healthcare team discuss exercise recommendations from the American Heart Association (AHA), American College of Cardiology (ACC), or Bethesda Guidelines? If so, which?

15. Which resources have you used to better understand your exercise recommendations?

- Letters or resources from doctor
- Support group
- Bethesda Guidelines
- AHA/ACC recommendations
- Family or friends
- Books
- Online websites
- Other (please specify)

16. How many minutes per week did you exercise prior to your diagnosis?

17. What activities or sports did you take part in before your diagnosis?

18. How many minutes per week do you exercise now after your diagnosis?

19. What activities or sports do you take part in now after your diagnosis?

20. Do you take any medication for your cardiac diagnosis, and if so which?

21. Did you have any additional medical procedures or surgeries following your diagnosis?

22. Do you have an Implantable Cardioverter Defibrillator (ICD)?

Yes

No

23. How many cardiac events have you had since your diagnosis? (Ex: syncope, cardiac arrest, arrhythmia)

24. If you have, was/were the cardiac event(s) during physical activity?

25. I wish that there were concrete or personalized recommendations for me to use for exercise

Yes

No

26. Describe how you were told about your exercise recommendations after your medical diagnosis?

27. How did you feel when you were informed about your medical diagnosis and exercise recommendations?

28. How do you feel now regarding your exercise recommendations?

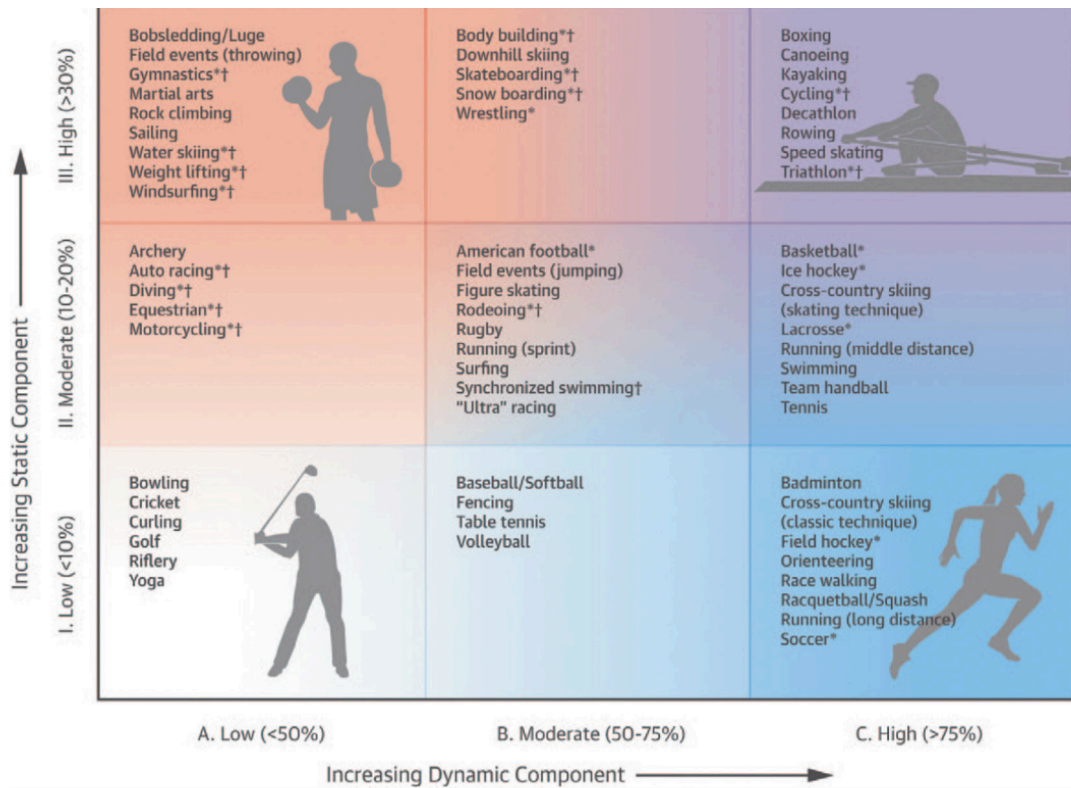
29. What are your motivations for either continuing or discontinuing exercise after your diagnosis?

Appendix C: Full ANOVA Table

Full ANOVA Table				
Comparison	df	F	n	p
Communication*Minutes	1	1.642	66	.205
Communication*Activity	1	4.327	66	.042**
Understanding*Minutes	1	2.024	65	.16
Understanding*Activity	1	4.541	65	.037**
Education*Minutes	1	4.434	65	.039**
Education*Activity	1	.086	65	.770
ICD*Minutes	1	3.256	66	.076
ICD*Activity	1	.069	67	.793

**=Statistically significant

Appendix D: 2015 Sport Classification Chart



Appendix E: Prevalence and Information on Genetic Diseases

Disease Name	Prevalence	Inheritance	Most Common Genes
Hypertrophic Cardiomyopathy (HCM)	1 in 500	Autosomal Dominant	<i>MYH7, MYBPC3, TTNT2, TTN3</i>
Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)	1 in 1,000- 1, 250	Autosomal Dominant	<i>PKP2</i>
Long QT Syndrome	1 in 2,000	Autosomal Dominant	<i>KCNE1, KCNE2, SCN5A</i>
Brugada Syndrome	1 in 2,000	Autosomal Dominant	<i>SCN5A</i>
Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT)	1 in 10,000	Autosomal Dominant	<i>RYR2, CALM1</i>
		Autosomal Recessive	<i>CASQ2, TRDN</i>