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I, Allyson Somers, hereby submit this original work as part of the requirements for the degree of Master of Science in Genetic Counseling.

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Provision of cardiovascular genetic counseling services: current practice and future directions

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Provision of cardiovascular genetic counseling services: current practice and future directions

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Abstract

Cardiovascular genetic counseling has emerged as a specialty critical to the care of patients with heritable cardiovascular disease. Although there is an increasing demand for genetic counseling services for cardiac disorders, the profession's strategies to meet this demand are not clear. As an initial, we sought to characterize the current practice patterns of cardiac genetic counseling by surveying the National Society of Genetic Counselors (NSGC) membership about cardiovascular training, education, and clinical practice related to cardiac disorders. Of the 105 (69%) participants who reported seeing a cardiac genetics patient in the previous 12 months, 42 (40%) identified themselves as a cardiovascular genetic counselor (CVGC) and 63 (60%) did not (non-CVGC). CVGCs were most likely to identify cardiology and non-CVGCs prenatal or pediatrics as their primary specialty. The most common conditions for which genetic counseling was provided were hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), long QT syndrome (LQTS), and genetic syndromes with cardiovascular disease. CVGCs were significantly more confident than non-CVGCs in providing genetic counseling for HCM, DCM, arrhythmogenic right ventricular cardiomyopathy, LQTS, Brugada syndrome, familial thoracic aortic aneurysm and dissection, and sudden cardiac death. While some graduate training programs provide learning opportunities for cardiovascular genetics, 86% of genetic counselors sought additional education related to cardiovascular genetics and listed online courses as the most desirable method of learning. Seventy-eight percent of non-CVGCs reported an interest in providing and need for cardiovascular genetic counseling services in their area. There is a growing need for and interest among the NSGC membership in cardiovascular genetic counseling. Additional training

resources and support for genetic counselors providing cardiovascular genetic counseling are needed.

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Introduction

Cardiovascular disease is the leading cause of death in the United States. Although environmental factors are known to contribute to cardiovascular disease risk, genetic factors also play an important role (Nabel, 2003). Over the past 20 years, major advances have been made in understanding the genetic etiology of several cardiovascular diseases including cardiomyopathy, congenital heart disease, thoracic aortic aneurysm and dissection, and arrhythmia (den Haan et al., 2009; Hofman, Tan, Alders, van Langen, & Wilde, 2010; Ingles, Yeates, & Semsarian, 2011; Kuzmik, Sang, & Elefteriades, 2012; Marino & Digilio, 2000; Pierpont et al., 2007). These advances in knowledge have been propelled in part by new technology for identifying DNA sequence variation. As such, clinical genetic testing for cardiovascular disease is increasingly available (Skrzynia, Demo, & Baxter, 2009). Most of these diseases demonstrate genetic and phenotypic heterogeneity and require specialized knowledge for genetic evaluation.

Numerous organizations and consensus statements recommend genetic testing and genetic counseling for inherited cardiovascular disease as part of standard clinical care (Ashley et al., 2012; Hershberger et al., 2009; Hiratzka et al., 2010). The American Heart Association (AHA) specifically recommends that expertise in cardiovascular genetics be utilized to direct the proper initiation, interpretation, and implementation of genetic testing (Ashley et al., 2012). In addition, the need for genetics professionals with cardiovascular expertise to provide comprehensive care for these patients and families has previously been reported (Demo, Skrzynia, & Baxter, 2009; Dunn, Caleshu, Cirino, Ho, & Ashley, 2013). These recommendations acknowledge the impact genetic counseling and testing have on both patients and their at-risk relatives. Genetic

counselors have unique and specific training in facilitating the understanding and adaption to the medical, psychological, and familial impact of genetic disease (Resta et al., 2006). Not surprisingly, the field has emerged as a specialty with the expertise necessary to evaluate patients with or at risk for genetic cardiovascular disease (Fowler, Napolitano, & Priori, 2009; Resta et al., 2006). Current recommendations for genetic counseling related to inherited cardiovascular disease include educating patients about the potential genetic etiology, inheritance patterns, recommendations for appropriate cardiac screening of at-risk relatives, and possible implications of clinical genetic testing. In the policy statement on genetics and cardiovascular disease, the AHA states that increased access to genetic counseling will be a key component to optimizing the benefits of clinical genetic testing (Ashley et al., 2012).

Despite the well established indication for genetic testing and counseling for patients with cardiovascular genetic disease, these services have not yet been integrated into the frontline of clinical cardiology (Fowler et al., 2009). One challenge may be the access to genetic counseling services. The 2012 National Society of Genetic Counselors (NSGC) Professional Status Survey (PSS) reports that only 24 (1.9%) genetic counselors identified cardiology as their primary specialty area. Additional literature regarding current practice and training of genetic counselors related to cardiovascular genetic counseling does not exist. Further defining the training, education, and scope of practice of genetic counselors providing services for cardiovascular disease is important to help prepare the profession to meet the growing demands of this patient population.

Accordingly, the objectives of this study were (1) to further characterize current cardiovascular genetic counseling practice by describing the genetic counselor work

setting, patient volume and cardiac disease-specific confidence and (2) to evaluate cardiac-specific sources of learning accessed by genetic counselors.

Methods

This study was approved by the Institutional Review Boards at Cincinnati Children's Hospital Medical Center and the University of Cincinnati College of Medicine. All participants were genetic counselors who were recruited through emails directed at the membership of the NSGC. Members received an email through the NSGC Listserv introducing the study and a link to the survey. The survey was available between 10/31/2012 and 11/28/2012 and a reminder email was sent 2 weeks after the initial invitation to participate. Participation in this survey was voluntary and all participants provided informed consent.

Instrumentation

A novel, quantitative survey was created in Research Electronic Database Capture (REDCap) hosted at (University of Cincinnati/Cincinnati Children's Hospital Medical Center) to assess the work settings, clinical activities, and experiences of genetic counselors providing cardiovascular genetic counseling (Appendix). Demographic questions were adapted from the NSGC PSS. The authors developed the remaining questions. The survey was comprised of 27 questions (20 multiple-choice, 6 yes/no, and 1 fill-in) and contained a skip pattern. The survey took approximately 10 minutes to complete. In addition to demographic information, participants were asked about their current work setting, clinical setting, number of cardiac patients and conditions seen,

confidence in providing cardiovascular genetic counseling, and sources of information and training.

The survey was piloted to genetic counselors in different specialties, including cardiovascular genetic counseling, at Cincinnati Children's Hospital Medical Center. After minor changes, the revised survey was dispensed to the NSGC Listserv.

Data Analysis

Descriptive statistics were used to summarize the clinical practice of the study population. Frequency/percentage was reported for categorical variables. Fisher's exact test was used to test equality of two proportions or to test for the independence/association of two nominal categorical variables. The Cochran-Armitage trend test was performed to assess for the presence of an association between a variable with two categories and an ordinal categorical variable. It tests for trends in binomial proportions across the levels of a single variable. Linear by linear association test was employed to look for any evidence of association between two ordered variables. The Cochran-Armitage trend test was used to compare the practice of cardiovascular genetic counselors (CVGCs) to genetic counselors who provided cardiovascular genetic counseling but did not identify cardiology as a specialty (non-CVGCs). To determine the confidence in providing genetic counseling for specific cardiovascular diseases, we examined the Likert-type scales using the Cochran-Armitage trend test. The analysis was performed in R (www.r-project.org).

Results

Practice Patterns

A total of 188 individuals responded to the survey. Thirty-five surveys were excluded due to partial completion, resulting in 153 completed surveys. Based on the membership of the NSGC at the time of the Professional Status Survey in 2012, the estimated response rate was 5%. Of the 105 participants who reported seeing a cardiac genetics patient in the previous 12 months, 42 (40%) identified themselves as a cardiovascular genetic counselor (CVGC) and 63 (60%) did not (non-CVGC). A higher percentage of CVGCs have combined, both clinical and non-clinical, roles in their position than non-CVGCs (31% vs. 13%, p value= 0.027). Forty-eight (31%) of the participants had not seen a cardiac genetics patient in the previous 12 months. Of those who had not seen a cardiac genetics patient, 10 were non-clinical and only demographic data were collected.

For this study, a cardiac genetics patient was defined as a patient referred for genetic counseling for a history of cardiovascular disease including cardiomyopathy, arrhythmia, vasculopathy/aortopathy, congenital heart disease, coronary heart disease, genetic syndrome with cardiovascular disease, or family history of sudden cardiac death (SCD). Cardiomyopathies included in the survey include hypertrophic cardiomyopathy (HCM), dilated cardiomyopathy (DCM), restrictive cardiomyopathy (RCM), arrhythmogenic right ventricular cardiomyopathy (ARVC), and left ventricular noncompaction (LVNC). Arrhythmias included long QT syndrome (LQTS) and Brugada syndrome (BrS). Genetic syndromes with cardiovascular disease included Alagille syndrome, Alstrom syndrome, Bardet-Biedl syndrome, Beckwith-Wiedemann syndrome,

CHARGE syndrome, chromosomal imbalance, Coffin-Lowry syndrome, Cornelia de Lange syndrome, dystrophinopathies (Duchenne/Becker muscular dystrophies), Ehlers-Danlos syndrome vascular type, Fabry disease, Friedreich's Ataxia, Holt-Oram syndrome, Leigh syndrome or other mitochondrial disorder, Loeys-Dietz syndrome, lysosomal storage disorder (Hunter syndrome, Hurler syndrome, Pompe disease, etc.), Marfan syndrome, myotonic dystrophy, Noonan syndrome, Russel-Silver syndrome, Smith-Magenis syndrome, Sotos syndrome, Shprintzen-Goldberg syndrome, Trisomy 13, Trisomy 18, Trisomy 21, Turner syndrome, Williams syndrome, and 22q11.2 deletion syndrome.

Differences were noted in primary specialty of counselors who identified as CVGCs versus those who do not. The majority of CVGCs identified their primary specialty as cardiology while none of the non-CVGCs identified their primary specialty as cardiology (66% vs. 0%, p value= 5×10^{-15}). Thirty-seven percent of non-CVGCs and only 7% of CVGCs identified their primary specialty as pediatrics (p value= 9×10^{-4}). Respondents were most likely to have provided cardiovascular genetic counseling for adult (78/104, 75%) and pediatric (60/104, 58%) patient populations. More CVGCs saw patients in conjunction with a cardiologist when seeing cardiac genetics patients compared to non-CVGCs (88% vs. 30%, p value= 3×10^{-9}); fewer CVGCs reported seeing cardiac genetics patients with a maternal fetal medicine specialist (7% vs. 27%, $p=0.01$). CVGCs were most likely to report having expertise for cardiomyopathy (41/42, 98%), arrhythmia (36/42, 86%), and vasculopathy/aortopathy (15/42, 36%). The characteristics of the participants are summarized in Table 1 and Table 2.

Cardiac Conditions

Overall, the most common conditions for which genetic counseling was provided in the previous 12 months were HCM (71%), DCM (61%), LQTS (56%), and genetic syndrome with cardiovascular disease (55%) (Table 3). Of the 56 respondents who reported providing counseling for a genetic syndrome, the most common genetic syndromes included Marfan syndrome (80%), Trisomy 21 (63%), and 22q11.2 deletion syndrome (57%). When stratified based on self-identification changes were noted in the types of patients for whom counseling was most frequent in CVGCs (n=42) when compared to non-CVGCs (n=63). Cardiomyopathies were the most frequently identified category for which CVGCs reported counseling, with HCM (88%), DCM (90%), and ARVC (86%) being the most frequently encountered. In addition, LQTS (83%) was a condition for which counseling was commonly provided. While those who did not identify as CVGCs also frequently counseled for HCM (60%), the most common conditions for which counseling was provided were congenital heart disease (65%) and genetic syndrome with a cardiac component (62%).

Confidence and Cardiovascular Genetic Counseling

Genetic counselor confidence in providing genetic counseling for specific diseases is summarized in Figures 1 and 2. Respondents were most likely to report being very confident when providing genetic counseling for HCM (65%, 48/74), LQTS, (61%, 36/59) BrS (70%, 23/33), and congenital heart disease (57%, 31/54) (Figure 1 and Figure 2). CVGCs were most likely to report being very confident when providing counseling for HCM (95%, 35/37), LQTS (77%, 27/35), BrS (91%, 20/22), and SCD (74%, 17/23)

(Figure 3 and Figure 4). The highest confidence among non-CVGCs was when providing counseling for congenital heart disease (59%, 24/41), coronary heart disease (40%, 2/5), HCM (35%, 13/37), and LQTS (38%, 9/24) (Figure 3 and Figure 4). Eight percent of CVGCs reported being somewhat unconfident in providing counseling for SCD (1/13) and four percent for congenital heart disease (1/23), while some non-CVGCs reported being either somewhat or very unconfident in providing counseling for 8 of 12 cardiac conditions or categories surveyed (HCM, DCM, LVNC, ARVC, coronary heart disease, LQTS, FTAAD, and SCD). This was highest for coronary heart disease, as 20% of non-CVGCs who provided counseling for this disease reported being unconfident. Those who identified as CVGCs were significantly more confident than those who do not identify as a CVGC for 7 of 12 conditions including: HCM, DCM, ARVC, LQTS, BrS, FTAAD, and SCD (Table 4).

Graduate and Professional Training

Of the respondents who had seen a cardiac genetics patient in the previous 12 months (n=105), 71% had cardiovascular disease lectures or presentations, 18% had a clinical cardiovascular rotation, and 4% were exposed to a cardiovascular genetics course during their graduate school training (Table 5). Twenty-eight percent were not exposed to any of the above during their graduate school training. Cardiovascular lectures were the most commonly reported training opportunities for both CVGCs and non-CVGCs. There was no significant influence of graduate training on counselors identifying as CVGCs (p-value ≥ 0.15).

Eighty-six percent (89/104) of respondents reported the need to obtain additional knowledge that was not taught in their genetic counseling program to provide cardiovascular genetic counseling. The majority of participants felt that an online course (82/139, 59%) would be most helpful in gaining knowledge of cardiovascular genetics. In the previous 12 months, 45% (62/137) of respondents reported attending a cardiology lecture at a national meeting.

Opinions about Cardiovascular Genetic Counseling

All CVGCs (41/41, 100%) reported an interest (agree or strongly agree) in providing cardiovascular genetic counseling services, while 78% (49/63) of non-CVGCs reported the same interest. The large majority of CVGCs (36/38, 95%) and 78% (49/63) of non-CVGCs reported (agree or strongly agree) a need for cardiovascular genetic counseling services in their city.

Discussion

Cardiovascular genetics is a growing field and genetic counseling and testing is recommended for many cardiac diseases. Genetic counselors have been identified as healthcare professionals with expertise needed to provide optimal care for patients and families with and at risk for genetic heart disease. We sought to characterize cardiovascular genetic counseling practice and identify cardiovascular genetics education and training accessed by members of the National Society of Genetic Counselors (NSGC). The 2012 NSGC Professional Status Survey (PSS) reports that 24 (1.9%) genetic counselors identified cardiology as their primary specialty area. Of the 105

(69%) participants who reported seeing a cardiac genetics patient in the previous 12 months, nearly twice as many (n=42, 40%) self-identified as a cardiovascular genetic counselor (CVGC) than in the PSS. Sixty-three (60%) respondents who did not identify as a CVGC (non-CVGC) reported providing cardiovascular genetic counseling in the previous 12 months. More CVGCs reported seeing patients with a cardiologist (n=37, 88%) than any other healthcare provider. CVGCs most commonly provided genetic counseling for HCM, DCM, ARVC, LQTS and SCD and were significantly more confident than non-CVGCs when providing services for 7 of 12 cardiac diseases. Seventy-four percent of genetic counselors were exposed to cardiovascular genetics education during graduate training, however, 86% reported the need for additional training in cardiovascular genetics and the majority indicated an online course as preferable.

Genetic Counselors and Cardiovascular Genetic Counseling

The majority of CVGCs (66%) reported cardiology as their primary specialty with a smaller group reporting cancer (12%). It is not unexpected that cancer genetic counselors may see cardiovascular genetics patients as both hereditary cancer and cardiovascular diseases are often adult onset. Respondents were most likely to provide cardiovascular genetic counseling for the adult patient population. Sixty-three (57%) respondents who did not identify as a CVGC had seen a cardiac genetics patient in the previous 12 months. This may be reflective of the increasing demand for cardiovascular genetic counseling services or an interest among a broader group of genetic counselors in cardiovascular genetics as the majority of respondents reported a perceived need for and

interest in providing cardiovascular genetic counseling services. While 100% (41/41) and 95% (36/38) of CVGCs reported an interest and need for cardiovascular genetic counseling services respectively, 78% (49/63) of non-CVGCs reported an interest and need for cardiovascular genetic counseling. Non-CVGCs who reported providing cardiovascular genetic counseling most often identified prenatal (30%) and pediatrics (37%) as their primary specialty. This is not unexpected as in the NSGC PSS, prenatal (29.2%) and pediatrics (13.5%) make up two of the three most commonly reported primary specialties of genetic counselors.

The majority of CVGCs saw patients with a cardiologist (88%) or geneticist (52%), while non-CVGCs were less likely to see patients with a cardiologist (30%). This survey did not address whether genetic counselors saw patients independently. The model of genetic counselors working with non-geneticist physicians has been well established in other specialties including maternal fetal medicine and oncology (Arnold & Self, 2012; Hemminki, Sundquist, & Lorenzo Bermejo, 2008). Michels et al. reported the methodology for genetic counseling, pre-symptomatic testing, and screening in the Netherlands involved the collaboration of a cardiologist, geneticist, and genetics nurse (Michels et al., 2007). In 2005, van Langen surveyed cardiologists and geneticists in regards to their preferences regarding each profession's role in providing genetic counseling and testing for HCM (van Langen et al., 2005). The results of this study indicated that both professions preferred a shared role; however, concerns were cited regarding capacity to see patients. Genetic counselors were not included in this survey. While a multi-disciplinary setting is likely ideal for care of patients with cardiovascular genetic disease, these clinics are often not accessible to the majority of individuals who

could benefit. It remains unclear of the optimal setting for genetic counselors to provide cardiovascular genetic counseling. Funding and other institutional infrastructure will likely play a large role in determining how genetic counseling services are integrated into the care of patients with cardiovascular genetic disease.

Of the thirteen cardiac conditions included in the survey, respondents were most likely to have provided counseling for HCM, DCM, LQTS, and genetic conditions with a cardiac disease component (most often Marfan syndrome, Trisomy 21, and 22q11.2 deletion syndrome). These conditions share in common the fact that they are well characterized and the majority are relatively common genetic conditions. Their clinical phenotypes are well established and they have highly accurate diagnostic tests. Finally, practice guidelines that include specific genetics recommendations exist for these conditions (Ackerman et al., 2011; Bassett et al., 2011; Bull, 2011; Hershberger et al., 2009; Keane & Pyeritz, 2008). In addition, CVGCs were likely to have provided counseling for ARVC (86%), a type of cardiomyopathy that is relatively common and for which genetic testing is incorporated into the diagnostic criteria (Marcus et al., 2010). Non-CVGCs were likely to have provided counseling for congenital heart disease (n=41, 65%), a common congenital anomaly that often presents in a prenatal or pediatric setting. CVGCs provided counseling more often for all of the specific cardiac genetics conditions and coronary heart disease, while non-CVGCs provided counseling more often for congenital heart disease or genetic conditions with a cardiac component. This is consistent with a CVGC's expertise for counseling for conditions where the cardiovascular aspect is the main component of a condition. Individuals with a genetic

condition are most often seen by a genetic counselor in conjunction with a clinical geneticist and may require management of extra-cardiac features.

Overall, respondents were most confident in providing genetic counseling for HCM, LQTS, BrS, and congenital heart disease. Some CVGCs expressed being unconfident in providing counseling only for sudden cardiac death and congenital heart disease, while non-CVGCs expressed being unconfident in providing counseling for 8 of 12 cardiac conditions. A smaller percentage of CVGCs (4-8%) reported being unconfident for specific conditions compared to non-CVGCs (up to 20%). Factors that may have influenced confidence include access to additional training opportunities, patient volume/counselor experience, and physician specialty involved. While the confidence in providing genetic counseling services for many diseases is quite high, this data identifies a need for additional learning opportunities for specific conditions including sudden cardiac death and congenital heart disease.

In addition to the low response rate of 5%, there were several limitations to this study. Forty percent of the respondents identified as a CVGC compared to 1.9% of respondents to the PSS. There was likely an ascertainment bias as there was a small number of non-CVGCs that completed the survey, so this subpopulation may not be reflective of the NSGC membership. Thus, the results may not accurately reflect the NSGC membership's cardiovascular genetic counseling practice patterns. The survey was designed to evaluate factors influencing confidence in providing cardiovascular genetic counseling as a whole but not for specific cardiac diseases. The definition of a cardiac genetics patient used in this study may be imperfect and may not encompass all cardiovascular diseases. For example, familial hypercholesterolemia was not included

and the survey did not include a free text option for other cardiovascular disease. In addition, the cardiac involvement for the genetic syndromes varied and there be some patients with a specific syndrome, like Marfan syndrome, who do not have cardiac involvement.

Cardiovascular Genetics Education and Training

Despite the fact that 74% of all respondents had some exposure to cardiovascular genetics (disease lectures or presentations, rotation, and/or course) during graduate training, the large majority (86%) reported the need to obtain additional knowledge to provide cardiovascular genetic counseling. The most common source of learning during graduate training was cardiovascular disease lectures or presentations. In addition to exposure to cardiovascular genetics to prepare counselors for the option for cardiology as their primary specialty, cardiovascular genetics is important knowledge for all genetic counselors. As cardiovascular genetics is a category on the board certification exam, learning opportunities on this topic are important for success and professional advancement. This indicates opportunities for graduate training programs to expand their curriculum to better integrate cardiovascular genetics. Interestingly, CVGCs were not more likely to have exposure to cardiovascular genetics education during their graduate training.

Learning opportunities are not limited to graduate training programs and professional development is an important component to a genetic counselor's career (Runyon, et al 2010). The most common sources of learning outside of graduate training reported were website/internet, medical journal and physicians. Over 50% of respondents

reported attending a conference/presentation and talking with a genetic counselor. In a survey of public health genetic counselors, the top two sources of learning were public health colleagues and going to a meeting (Powell, Hasegawa, & McWalter, 2010). The sources of learning were similar between public health genetic counselors and the respondents in this study, but the percentages were higher overall for sources of learning sought by counselors providing cardiovascular genetic counseling.

The majority of participants felt that an online course would be the most helpful learning opportunity in gaining knowledge of cardiovascular genetics. Currently, category 1 continuing education units (CEUs) offerings through the NSGC related to cardiovascular genetics include the NSGC Cardiovascular Special Interest Group case conference and an online Cardiovascular Genetics Course offered by the University of Cincinnati/Cincinnati Children's Hospital Genetic Counseling Program. Between 1/2012 and 3/2013, 72 genetic counselors have taken the course.

Future Directions

Given the wide variety of cardiovascular genetic diseases and associated medical management issues, genetic counselors need to engage both geneticists and cardiologists in providing cardiovascular genetic counseling. While geneticists may be familiar with a genetic counselor's training and scope of practice, cardiologists may need additional education on recommendations for genetic counseling and testing and the benefit of incorporating genetic counselors into patient care. With the increasing understanding of the genetic aspects of cardiovascular conditions and clinical testing, cardiologists may also need additional training on the genetics of cardiovascular conditions and the benefits

of collaborating with genetic counselors in providing care (den Haan et al., 2009; Ingles et al., 2011; Marino & Digilio, 2000; Pierpont et al., 2007; Skrzynia et al., 2009).

Conclusions

Cardiovascular genetic counseling is an emerging subspecialty with an increasing number of both CVGCs and non-CVGCs providing cardiovascular genetic counseling. Cardiology was CVGCs primary specialty and adult the primary patient population. CVGCs were more confident in providing counseling for the majority of cardiovascular conditions. While some graduate training programs provide cardiovascular genetic training, most genetic counselors sought additional education and cited online courses as the most desirable method of learning. Additional training resources and support for genetic counselors providing cardiovascular genetic counseling are needed.

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Table 1 Self-reported Practice Patterns of Surveyed Genetic Counselors

	CVGCs	Non-CVGCs	p-value
Position	n=42	n=63	
Clinical	29 (69%)	55 (87%)	0.03
Clinical and non-clinical	13 (31%)	8 (13%)	
Specialty	n=41	n=63	
Prenatal	2 (5%)	19 (30%)	0.002
Pediatrics	3 (7%)	23 (37%)	9.0×10^{-4}
Other	0 (0%)	10 (16%)	0.006
Lab	1 (2%)	1 (2%)	1.0
Cancer	5 (12%)	5 (8%)	0.51
Adult	3 (7%)	5 (8%)	1.0
Cardiology	27 (66%)	0 (0%)	5.0×10^{-15}
Physician	n=42	n=63	
Cardiologist	37 (88%)	19 (30%)	3.0×10^{-9}
Geneticist	22 (52%)	43 (68%)	0.11
Obstetrician	1 (2%)	6 (10%)	0.24
Maternal Fetal Medicine	3 (7%)	17 (27%)	0.01
Other	4 (10%)	5 (8%)	1.0

Note: CVGCs are respondents who identify as cardiovascular genetic counselors. Non-CVGCs are respondents who do not identify as cardiovascular genetic counselors.

Table 2 CVGCs Self-reported Expertise of Types of Cardiovascular Disease

Types of Disease	n=42
Cardiomyopathy	41 (98%)
Arrhythmia	36 (86%)
Vasculopathy	15 (36%)
Congenital heart disease	11 (26%)
Genetic syndromes with cardiac disease	12 (29%)
Other	1 (2.4%)

Table 3 Genetic Counseling Services Provided for Cardiovascular Conditions

Condition	CVGCs n=42	Non-CVGCs n=63	All n=105
HCM	37 (88%)	38 (60%)	75 (71%)
DCM	38 (90%)	26 (41%)	64 (61%)
LVNC	24 (57%)	7 (11%)	31 (30%)
ARVC	36 (86%)	9 (14%)	45 (43%)
RCM	10 (24%)	2 (3%)	12 (11%)
Congenital heart disease	13 (31%)	41 (65%)	54 (51%)
Coronary heart disease	9 (21%)	5 (8%)	14 (13%)
LQTS	35 (83%)	24 (38%)	59 (56%)
BrS	22 (52%)	11 (17%)	33 (31%)
FTAAD	22 (52%)	19 (30%)	41 (39%)
Vasculopathy/ Aortopathy	16 (38%)	10 (16%)	26 (25%)
SCD	33 (79%)	17 (27%)	50 (48%)
Genetic syndrome	19 (45%)	39 (62%)	58 (55%)

Table 4 CVGC Status and Influence on Confidence in Providing Cardiovascular Genetic Counseling

Condition:	p value
HCM	2.30 x 10⁻⁶
DCM	0.004
LVNC	0.1
ARVC	0.002
RCM	0.06
Congenital heart disease	0.13
Coronary heart disease	0.87
LQTS	0.002
BrS	1.80 x 10⁻⁴
FTAAD	0.021
Vasculopathy/Aortopathy	0.11
SCD	0.01

Note: Significant p values (p< .05) are bolded.

Table 5 Reported Cardiovascular Genetics Training Opportunities

Graduate Training Opportunity	CVGCs n=42	Non-CVGCs n=63	All n=105
Cardiovascular Genetics Course	0 (0%)	4 (6%)	4 (4%)
Cardiovascular lectures	27 (63%)	48 (76%)	75 (71%)
Clinical cardiovascular rotation	7 (9%)	12 (19%)	19 (18%)
None	15 (35%)	14 (22%)	29 (28%)

Figure 1. Genetic counselors' overall confidence in providing genetic counseling for cardiomyopathies. (HCM, n=74; DCM, n=61; LVNC, n=32; ARVC, n=44; RCM, n=11).

Figure 2. Genetic counselors' overall confidence in providing genetic counseling for additional cardiovascular conditions. (Congenital heart disease, n=54; coronary heart disease, n=12; LQTS, n=59; BrS, n=33; FTAAD, n=41; vasculopathy/aortopathy, n=26; SCD, n=40).

Figure 3. Genetic counselors' overall confidence in providing genetic counseling for cardiomyopathies based on CVGC status. (HCM, n=74; DCM, n=61; LVNC, n=32; ARVC, n=44; RCM, n=11).

Figure 4. Genetic counselors' overall confidence in providing genetic counseling for additional cardiovascular conditions based on CVGC status. (Congenital heart disease, n=54; coronary heart disease, n=12; LQTS, n=59; BrS, n=33; FTAAD, n=41; vasculopathy/aortopathy, n=26; SCD, n=40).

Appendix

Questionnaire

Demographics questions

1. What is your gender?
 - ☐ Male
 - ☐ Female
2. What is your race?
 - ☐ Asian
 - ☐ Black or African American
 - ☐ White or Caucasian
 - ☐ Native Hawaiian or Other Pacific Islander
 - ☐ American Indian or Alaskan Native
 - ☐ Other
3. What is your ethnicity?
 - ☐ Hispanic
 - ☐ Non-Hispanic
 - ☐ Unknown
4. How old are you?
5. What year did you graduate from a genetic counseling/medical genetics graduate program?
 - ☐ 2010 - 2012
 - ☐ 2005-2009
 - ☐ 2000-2004
 - ☐ 1995-1999
 - ☐ 1990-1994
 - ☐ 1985-1989
 - ☐ 1980-1984
 - ☐ 1975-1979
 - ☐ 1970-1974
6. What is your work status?
 - ☐ Full time
 - ☐ Part time
7. In what NSGC region do you practice genetic counseling?
 - ☐ Region 1 (CT, MA, ME, NH, RI, VT, Canadian Maritime Provinces)
 - ☐ Region 2 (DC, DE, MD, NJ, NY, PA, VA, WV, Quebec, Puerto Rico, Virgin Islands)
 - ☐ Region 3 (AL, FL, GA, KY, LA, MS, NC, SC, TN)
 - ☐ Region 4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario)

- Region 5 (AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Saskatchewan)
- Region 6 (AK, CA, HI, ID, NV, OR, WA, British Columbia)

8. In what type of community do you practice genetic counseling?

- Urban
- Rural
- Unknown

9. Are cardiovascular genetics services available in your city?

- Yes
- No
- Unsure

Work setting questions

10. What is your primary work setting?

- University Medical Center
- Private Hospital/Medical Facility
- Public Hospital/Medical Facility
- Physician's Private Practice
- Diagnostic Laboratory
- Genetic Counseling Private Practice
- Other (please specify): _____

11. What is your primary specialty area?

- Prenatal
- Cancer
- Pediatric
- Adult
- Cardiology
- Research
- Laboratory
- Other (please specify): _____

12. Is your current position:

- Clinical
- Non-clinical
- Both
 - If Non-clinical is checked, thank you for your participation in the study.

Clinical questions

13. Have you seen a cardiac genetics patient in the past 12 months? (A cardiac genetics patient is defined as a patient referred for genetic counseling for a history of cardiovascular disease including cardiomyopathy, arrhythmia, vasculopathy/aortopathy, congenital heart disease, coronary heart disease, genetic syndrome with cardiovascular

disease, or family history of sudden cardiac death.)

- ☐ Yes
- ☐ No (go to question 21a)

14. Is 50% or more of your caseload attributed to counseling for cardiac disease?

- ☐ Yes
- ☐ No

15a. Do you identify yourself as a cardiac genetic counselor?

- ☐ Yes
- ☐ No

IF YES,

15b. How many cardiac genetics patients have you seen in clinic in the past month?

- ☐ 1
- ☐ 2-5
- ☐ 6-10
- ☐ 11-15
- ☐ 16-20
- ☐ 21-25
- ☐ >25

15c. For what types of disease do you have expertise?

- ☐ Cardiomyopathy
- ☐ Arrhythmia
- ☐ Vasculopathy/Aortopathy
- ☐ Congenital heart disease
- ☐ Coronary heart disease
- ☐ Genetic syndromes with cardiac disease
- ☐ Other (please specify): _____

IF NO,

15d. How many cardiac genetics patients have you seen in clinic in the past 12 months?

- ☐ 1
- ☐ 2-5
- ☐ 6-10
- ☐ 11-15
- ☐ 16-20
- ☐ 21-25
- ☐ >25

16. For what patient population have you seen cardiac genetics patients in the previous 12 months? (check all that apply)

(A cardiac genetics patient is defined as a patient referred for genetic counseling for a history of cardiovascular disease including cardiomyopathy, arrhythmia,

vasculopathy/aortopathy, congenital heart disease, coronary heart disease, genetic syndrome, or family history of sudden cardiac death.)

- ☐ Adult
- ☐ Pediatric
- ☐ Prenatal
- ☐ Other (please specify): _____

17. For what clinical setting have you seen cardiac genetics patients in the previous 12 months? (check all that apply)

- ☐ General pediatrics clinic
- ☐ Adult clinic
- ☐ Prenatal clinic
- ☐ Multidisciplinary clinic
- ☐ Cardiology clinic
- ☐ Other (please specify): _____

18. What other healthcare professionals were involved during the session with the cardiac patients you have seen in the previous 12 months? (check all that apply)

- ☐ Cardiologist
- ☐ Geneticist
- ☐ Obstetrician
- ☐ Maternal Fetal Medicine
- ☐ Other (please specify): _____

19a-b. For which of the following conditions have you provided cardiac genetic counseling for in the previous 12 months? (check all that apply)

Condition		Provided Counseling (Y/N)
Hypertrophic cardiomyopathy		
Dilated cardiomyopathy		
Left ventricular non-compaction		
Arrhythmogenic right ventricular cardiomyopathy		
Restrictive cardiomyopathy		
Congenital heart disease (structural heart disease)		
Coronary heart disease		
Long-QT syndrome		
Brugada syndrome		
Familial thoracic aortic aneurysm and dissection (FTAAD)		
Vasculopathy/Aortopathy		
Family history of sudden cardiac death (SCD)		
Other (please specify): _____		
Genetic condition with cardiac disease	Alagille syndrome	
	Alstrom syndrome	
	Bardet-Biedl syndrome	
	Beckwith-Wiedemann syndrome	

	CHARGE syndrome	
	Chromosomal imbalance	
	Coffin-Lowry syndrome	
	Cornelia de Lange syndrome	
	Dystrophinopathies (Duchenne/ Becker muscular dystrophies)	
	Ehlers-Danlos syndrome, vascular type	
	Fabry disease	
	Friedreich's Ataxia	
	Holt-Oram syndrome	
	Leigh syndrome or other mitochondrial disorder	
	Loeys-Dietz syndrome	
	Lysosomal storage disorder (Hunter syndrome, Hurler syndrome, Pompe disease, etc.)	
	Marfan syndrome	
	Myotonic dystrophy	
	Noonan syndrome, LEOPARD, or other RASopathy	
	Russel-Silver syndrome	
	Smith-Magenis syndrome	
	Sotos syndrome	
	Shprintzen-Goldberg syndrome	
	Trisomy 13	
	Trisomy 18	
	Trisomy 21	
	Turner syndrome	
	Williams syndrome	

20. How confident were you in providing counseling for the specific condition(s)?

- Only those checked in 19a will appear for participants.

	Very Confident	Somewhat Confident	Neutral	Somewhat Unconfident	Very Unconfident
Hypertrophic cardiomyopathy					
Dilated cardiomyopathy					
Left ventricular non-compaction					
Arrhythmogenic right ventricular cardiomyopathy					
Restrictive cardiomyopathy					
Congenital heart disease					

(structural heart disease)					
Coronary heart disease					
Long-QT syndrome					
Brugada syndrome					
Familial thoracic aortic aneurysm and dissection (FTAAD)					
Vasculopathy/Aortopathy					
Family history of sudden cardiac death					

Non-clinical questions

21. Did you feel you needed to obtain additional knowledge that was not taught in your genetic counseling training program to provide cardiac genetic counseling?

- ☐ Yes
- ☐ No

22a. Have you ever referred a patient to a geneticist or genetic counselor who specializes in cardiovascular disease?

- ☐ Yes
- ☐ No
- ☐ Not applicable

22b. For what type(s) of disease did you refer one or more patients to a geneticist or genetic counselor who specializes in cardiovascular disease? (check all that apply)

- ☐ Cardiomyopathy
- ☐ Congenital heart disease
- ☐ Coronary heart disease
- ☐ Inherited arrhythmia
- ☐ Vasculopathy/Aortopathy
- ☐ Genetic syndrome
- ☐ Other (please specify): _____

23. What sources of information have you used to obtain up-to-date genetic information regarding cardiovascular disease? (check all that apply)

- ☐ Website/internet
- ☐ Medical journal
- ☐ Clinical genetic counselors
- ☐ Laboratory genetic counselors
- ☐ Physicians
- ☐ Textbooks
- ☐ Online cardiovascular genetics course
- ☐ Conference/presentation (please specify): _____
- ☐ Cardiovascular NSGC Special Interest Group (SIG)
- ☐ None

- Other (please specify): _____

24. Were any of the following incorporated to your graduate school training? (check all that apply)

- Cardiovascular genetics course
- Cardiovascular disease specific lectures/presentations
- Clinical cardiovascular rotation
- None
- Other (please specify): _____

25. In the past 12 months, which of the following have you done? (check all that apply)

- Took online course on cardiology
- Attended local meeting related to cardiology
- Attended national meeting related to cardiology
- Attended cardiology lecture at national meeting
- Consulted Cardiovascular NSGC SIG
- None

26. Which of the following do you feel would be MOST helpful in gaining knowledge of cardiovascular genetics?

- Online course on cardiology
- Local meeting related to cardiology
- National meeting related to cardiology
- Cardiology lecture at national meeting
- Consulted Cardiovascular NSGC SIG
- None

27a-i. Please state how strongly you agree with the following statements:

	Strongly Agree	Agree	Unsure	Disagree	Strongly Disagree
I am interested in providing genetic counseling for cardiovascular disease.					
There is a need for cardiovascular genetics services in my city.					
There is a need for additional genetic counselors with cardiovascular expertise in my city.					
There is a need for additional cardiologists with cardiovascular expertise in my city.					
There is a need for additional geneticists with cardiovascular expertise in my city.					
Cardiologists in my city are interested in genetic testing for patients with cardiovascular disease.					
Geneticists in my city are interested in genetic testing for patients with cardiovascular disease.					

There is a need for cardiovascular genetics lectures during graduate school training.					
There is a need for cardiovascular genetics clinical rotations during graduate school training.					