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A Review of the Emergence and Expansion of Cardiovascular Genetic Counseling

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Abstract

Purpose of Review The genetic counseling profession is growing rapidly, as is its presence in cardiology. In this review, we will survey recent innovations and research in cardiovascular genetic counseling, as well as findings from the broader genetic counseling field that are relevant to cardiovascular genetic counseling.

Recent Findings Research into the structure of services finds that genetic counselors are increasingly being embedded within cardiology departments and that cardiologists value their expertise and contributions. Findings from other genetic counseling subspecialties and from clinical psychology suggest this trend toward moving genetic counselors into the cardiology clinic will increase access, timeliness, and uptake of genetics services. Studies on the selection and interpretation of genetic tests in cardiology have shown that there is a need for specialized expertise in these areas. This has led clinical genetic counselors, in collaboration with their physician colleagues, to take a more active role in assessing whether genetic variants contribute to disease. Evidence on the psychological and behavioral impact of cardiovascular genetic counseling is beginning to emerge with several non-randomized and one randomized study showing benefit. As investigators, genetic counselors are leading studies that generate practice-shaping insights into inherited cardiovascular disease, including the genetic underpinnings of disease, natural history, prognosis, psychological impact, and genetic test interpretation.

Summary Genetic counseling is in its early days as an academic and scientific discipline, yet there are, nonetheless, several notable recent findings that are shaping the field.

Keywords Genetic counseling · Cardiac genetics · Interdisciplinary care · Genetic testing · Service delivery · Cardiovascular genetic counseling

Introduction

To set the context for consideration of recent developments in cardiovascular genetic counseling, we first review the landscape of genetic counseling practice and research.

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Genetic counseling is both a clinical intervention and a profession. As a clinical intervention, genetic counseling has been defined by the National Society of Genetic Counselors as a "process of helping people understand and adapt to the medical, psychological and familial implications of genetic contribution to disease," [1]. This process encompasses education and counseling, which is aimed at helping patients make informed decisions and assisting them in psychological and behavioral adaptation to genetic disease or risk [2].

The genetic counseling profession in the USA is made up of approximately 5000 professionals with diverse roles in clinics, genetic testing laboratories, research, and education. Genetic counselors have Masters degrees from accredited genetic counseling training programs and are board-certified and licensed. Training includes clinical experience in many medical specialties and development of expertise in genomics, education, family history assessment, genetic test selection and interpretation, and focused psychological and behavioral counseling [3, 4].

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As a whole, the genetic counseling profession has been growing rapidly (Fig. 1). The number of practicing genetic counselors doubled between 2006 and 2018 and is expected to grow by another 29% by 2026 [5, 6]. On the backdrop of that growth, there has also been a rapid increase in the number of genetic counselors providing care to families with inherited cardiovascular disease (Fig. 1).

Genetic counselors first began to specialize in cardiology in the early 2000s, when genetic testing for cardiovascular disease became available [7, 8]. In the decade that followed, there was a rapid increase in available genetic tests for inherited cardiovascular conditions, with a parallel increase in the number of genetic counselors specializing in cardiology. By 2018, 12% of genetic counselors were providing cardiovascular genetics care [3] (Fig. 1). Multiple cardiovascular professional societies have now published guidelines that recommend genetics evaluations for cardiovascular disorders, which includes genetic counseling [9–15].

Genetic counseling has been a primarily clinically oriented profession, and a relatively young one at that, with the first genetic counseling class graduating in 1971. The field is in its early days as an investigative academic discipline, with only a minority of genetic counselors functioning as investigators. The field will presumably develop a robust research agenda that drives evidence-based practice, as much as clinical psychology, social work, and nursing have [22]. Indicative of how early genetic counseling is in its development as an academic discipline, a recent meta-analysis of randomized control trials in genetic counseling identified just 58 studies [23•]. Nonetheless, there are recent innovations and research that are notable and are shaping practice. We will review several such studies from cardiovascular genetic counseling specifically, as well as applicable research from the genetic counseling field more broadly.

Embedding Genetic Counselors in the Cardiology Clinic

Perhaps the most significant and impactful innovation in cardiovascular genetic counseling has been the embedding of specialized genetic counselors in cardiology clinics. Historically, genetics services were provided by genetic counselors and/or geneticists who did not specialize in cardiology and were located in a separate department or even a separate hospital. In the early 2000s, genetic counselors started to move into the cardiology clinic, providing care alongside cardiologists and developing expertise in cardiovascular genetics [8, 24–26]. Several studies have documented this innovation.

Spoonamore and colleagues surveyed cardiologists who see patients with inherited cardiovascular disease [24]. Of the cardiologists whose patients are cared for by a genetic counselor, 75% had a genetic counselor within their department. This practice of embedding genetic counselors in cardiology had been occurring for a mean of 6.4 years. Physician-scientists who study genetics were the earliest to embed genetic counselors in cardiology; they had worked with a genetic counselor for twice as long and had more genetic counselors in their department. The study also found that the trend of embedded genetic counselors has begun expanding beyond these early-adopter physicians who study genetics. Interviews revealed that cardiologists highly value genetic counselors and appreciate the rigor of their training and the expertise genetic counselors have developed in cardiovascular genetics.

Erskine and colleagues describe one example of embedding genetic counselors within an interdisciplinary team in cardiology [25]. This team consists of clinicians specializing in clinical genetics, genetic counseling, pediatric and adult cardiology, nursing, psychology, and ethics. They describe

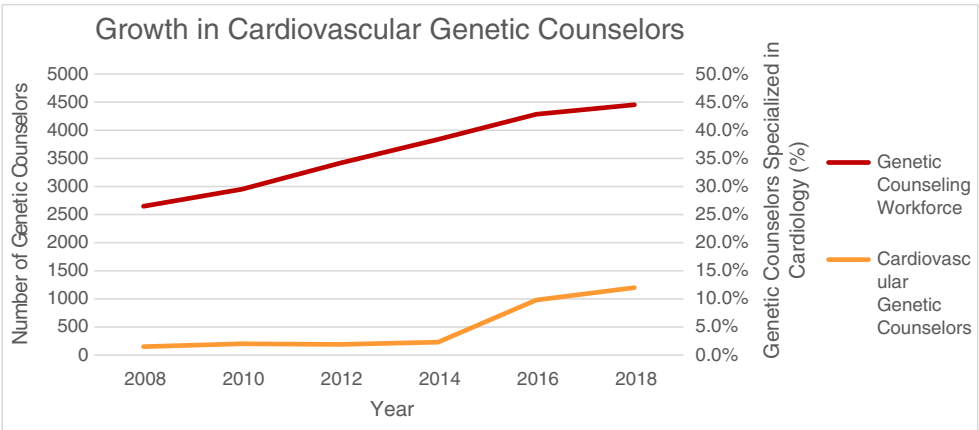

Fig. 1 Growth in cardiovascular genetic counselors. Red line represents the total number of certified genetic counselors per the American Board of Genetic Counseling [16]. Orange line represents the percentage of genetic counselors who report cardiology as a specialty in the National Society of Genetic Counselors professional status survey [3, 17–21]. Due to differences in reporting, data from 2008 to 2014 represent the number of genetic counselors who spend 50% or more of their time seeing cardiology patients. Data from 2016 and 2018 represent the number of genetic counselors who have spent *any time* seeing patients with cardiovascular indications.

several illustrative cases that demonstrate how each team member's expertise contributes to the care of the patient in an interactive and collaborative fashion. Similar models have been described by other groups in the USA and internationally [8, 27].

Indicative of the need for the collaborative model described by Erskine and colleagues, a study in the Netherlands found that cardiologists feel responsible for discussing a diagnosis of hypertrophic cardiomyopathy (HCM) and its hereditary nature, but would prefer that genetics specialists handle coordinating clinical and genetic screening for family members [28]. In this study, both cardiologists and clinical geneticists preferred a collaborative model for care of these patients. Helm and colleagues provide insight into the volume of genetic counseling in cardiology clinics. They embedded a genetic counselor in a pediatric arrhythmia clinic and found that 14% of patients needed genetic counseling services [29•].

The collaborative nature of interdisciplinary care in cardiovascular genetics is demonstrated in Reuter and colleagues' study on genetic test interpretation practices [30•]. They found this to be an interdisciplinary activity, with the majority of genetic counselors (81%) reporting they interpret genetic tests in collaboration with cardiologist and/or geneticist colleagues. Such close collaboration is facilitated by the genetic counselor being housed in cardiology. Indicative of the expertise genetic counselors bring to the team, they found that the genetic counselor is nearly always the team member primarily responsible for gathering evidence to inform the genetic test interpretation (95%). In three-quarters of teams, the genetic counselor was also the team member primarily responsible for the interpretation of the genetic test.

The maturation of this trend toward interdisciplinary care teams with genetic counselors embedded in cardiology is evident in the American Heart Association's recent recommendation that an interdisciplinary care team including genetic counselors be involved in the clinical care of patients with inherited cardiovascular diseases [31].

Looking elsewhere in the genetic counseling literature, several studies in cancer genetics have found that embedding genetic counselors in specialty medical clinics improves access [32–35]. For example, Senter and colleagues found a significant increase in the proportion of patients who received genetic counseling from the referral model (38%) to the embedded model (82%), as well as a reduction in wait time [33]. They attributed this increase to reducing the number of steps required to schedule an appointment. Each step is another opportunity to lose the patient to follow-up and takes time the patient may not have. Similar findings have been seen in behavioral medicine, with embedding psychologists in medical clinics increasing access to and uptake of services [36, 37]. Pedersen et al. found that embedding genetic counselors in oncology clinics not only increased access, it also impacted management. They observed a 69% increase in the number of patients who had genetic testing in time to make key surgical decisions, decisions that depend on the results of such tests [35]. Anecdotally, in our own center, we have seen that both embedded and on-call genetic counseling allow for timely genetics evaluations that can inform critical treatment decisions such as whether to implant a defibrillator or perform aortic root replacement. Studies have yet to be done on the impact of embedding genetic counselors in cardiology clinics.

Genetic Test Selection and Interpretation

Genetic testing has been found to be cost-effective for many inherited cardiovascular diseases and multiple guidelines recommend genetic testing [8–15, 38–40]. However, cost-effectiveness and benefit both hinge on the correct implementation of genetic testing. Recent studies have revealed the complexity of genetic testing and how genetic counselors can help ensure that genetic tests are interpreted and used appropriately.

A case series by Arscott and colleagues demonstrates that the involvement of a genetic counselor reduces the cost of genetic testing by: (1) choosing the most appropriate genetic test, (2) identifying the best person in the family to test, and (3) interpreting results accurately to facilitate appropriate medical management [41]. Studies have also found that having genetic counselors review genetic test orders reduces costs by modifying or canceling inappropriate orders [42–46]. In two such studies, about half of ordered genetic tests were canceled due to inappropriate test selection or limited clinical benefit to the patient, saving more than $2000 per patient [43, 46].

Genetic counselors in the cardiology clinic have increasingly played a key role in interpreting the results of genetic tests, as evidenced by the findings of Reuter and colleagues [30•]. Nearly all of the clinical cardiovascular genetic counselors (96%) they surveyed gather additional information on variants identified on test reports and most (81%) make an independent assessment regarding pathogenicity. Their data show that variant classification is a relatively recent (mean 3.2 years) addition to clinical practice. Prior to this shift, the primary locus of responsibility for the interpretation of genetic tests lay with genetic testing laboratories. One of the driving forces behind clinical cardiovascular genetic counselors and their teams taking greater responsibility for test interpretation has been evidence that laboratories and clinicians often disagree on test interpretation [30•,47–49].

Another reason that cardiovascular genetic counselors started taking a more active role in genetic test interpretation was their experience of the impact of misinterpretation on families [30•]. Arscott and colleagues report an illustrative case: a healthy male underwent genetic testing after his sibling died suddenly [41]. A variant in a long QT syndrome gene was

identified and the laboratory described it as a "probable disease-causing mutation." Over 20 relatives underwent genetic testing for this variant, some of whom had implantable cardioverter defibrillators placed based solely on positive genetic test results. The patient was then referred to an interdisciplinary cardiovascular genetics clinic. After gathering data on the variant, the genetic counselor determined that there was insufficient evidence to support the variant as the cause of the brother's sudden death. Clinical data revealed that many family members who carried the variant did not have a prolonged QT, further calling into question the role of the variant in disease. In the case of family members who have the variant, many were diagnosed and treated inappropriately, while those who did not were prematurely released from cardiac screening. Unfortunately, cases such as these are not uncommon [50, 51].

To combat these issues, the American College of Medical Genetics developed updated criteria for genetic testing interpretation [52]. However, these guidelines are not gene- or disease-specific. This lack of specificity has been shown to negatively impact the classification of variants in cardiovascular genes [53•]. The Clinical Genome Resource (ClinGen) seeks to create gene-specific guidelines to remedy this issue. The first of these guidelines to be published was in 2018 for *MYH7* and hypertrophic cardiomyopathy [53•]. Gene- and disease-specific guidelines are also now available for *LDLR* [54] with guidance for additional cardiovascular diseases in development. ClinGen has also been working to assess the strength of evidence behind gene-disease associations; they found that two-thirds of genes with a reported association with HCM have limited or no evidence to support their role in disease [55]. These studies are shifting the way cardiovascular genetic counselors select and interpret genetic tests.

Recent studies have also revealed the critical need for adjudication of some applications of the consumer-initiated genetic tests provided by companies like 23andMe and Ancestry.com [56, 57]. Consumers can download their raw, uninterpreted data from these companies' websites. These data are beyond what is included in reports provided by these companies, have not undergone quality control, and contain variants that the underlying assays were not optimized to detect. Consumers can then upload these data to third-party websites that assert pathogenicity of variants via algorithms that are not clinically validated. In this way, some people learn they are predisposed to inherited cardiovascular disease and undergo extensive evaluations, only to later find out that the finding was a false positive [56]. One clinical laboratory tested 49 patients for variants identified via such methods and found that 40% of them were false positives, underscoring the importance of confirmatory genetic testing [57]. The work of adjudicating such results, educating patients, and confirming the results is increasingly falling to cardiovascular genetic counselors [56].

Family History Collection and Assessment

Assessment of a thorough three-to-four-generation pedigree is recommended in multiple guidelines as a core part of the care of families with inherited cardiovascular disease [9–15]. This aspect of care is often accomplished by cardiovascular genetic counselors, who have extensive training in family history collection and assessment.

Waddell-Smith and colleagues found that collection of family history by a genetics specialist significantly increased sensitivity [58]. More than half of the 37 inpatients with a suspected inherited cardiovascular disease studied did not have their family history recorded prior to evaluation by a genetics specialist. When family history had been collected, it was insufficient to accurately assess for familial disease. Family history assessment by a genetics nurse led to a diagnosis of a previously missed familial disease in 41% of cases.

Obtaining thorough family histories is time-consuming. Waddell-Smith and colleagues noted it took between 30 and 90 min [58]. To increase efficiency, some genetic clinics are moving to initial collection of family history via online tools [59]. Other clinics obtain family history information over the phone prior to the in-person appointment, which one study indicates may increase patients' self-efficacy [60].

Education and Counseling

Genetic counseling has been described as a "highly circumscribed form of psychotherapy," in which counseling techniques and personalized information are used to help the patient adapt to their disease and to promote behavior change [61]. Several recent studies have shed light on the impact of cardiovascular genetic counseling on psychological and behavioral outcomes.

Caleshu and colleagues describe four cases in which patients with inherited cardiovascular disease benefited from psychological care provided by genetic counselors embedded in the cardiovascular clinic (Fig. 2) [27]. Genetic counselors employed empathic and supportive counseling, psychoeducation, and normalization to help these patients to (1) adjust to their diagnosis, exercise restrictions, and ICD shocks, (2) make a decision about ICD implantation, (3) grieve the sudden death of a family member, and (4) adjust to life after cardiac arrest in a child.

Going beyond case series, several recent non-randomized studies have shown psychological benefit from cardiovascular genetic counseling. In a single-center study of 48 adult patients, cardiovascular genetic counseling increased patients' awareness of family

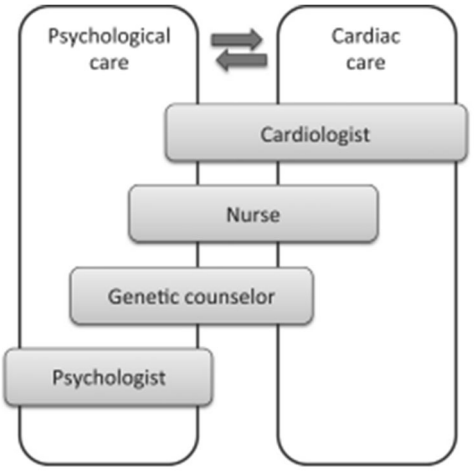

Fig – Interdisciplinary psychological and medical care for inherited cardiovascular diseases. There is a bidirectional relationship between psychological care and medical care with each domain impacting the other. Each member of the interdisciplinary team contributes to the comprehensive care of the patient and family. The psychologist contributes primarily to the psychological care of the patient. The cardiologist, nurse, and genetic counselor each contribute to both the psychological and cardiac care, though to different extents depending on each team member's role.

Fig. 2 Team roles in interdisciplinary care for inherited cardiovascular disease. The figure illustrates the division of psychological care and medical care among members of the interdisciplinary team. With permission from Caleshu et al. [27]

screening recommendations from 40 to 76% [62]. There was also a sizable improvement in scores on the Genetic Counseling Outcome Scale (Cohen's $d = 0.94$). This measure was designed to assess the impact of genetics services. It captures key psychological, educational, and behavioral outcomes such as understanding the genetic condition, a sense of control, emotional impact, and a sense of self-efficacy [63]. Using a Dutch translation of the Genetic Counseling Outcome Scale, Voorwinden and colleagues also saw improvement after genetic counseling in a large sample of 2194 patients from two centers, 21% of whom presented for care because of cardiovascular disease in the family (Cohen's $d = 0.30$) [64]. In a single-center study of patients with arrhythmogenic cardiomyopathy, Murray and colleagues observed significant improvement in the Genetic Counseling Outcome Scale pre- and post-cardiovascular genetic counseling ($p < 0.0001$), as well as a decrease in cardiac anxiety ($p = 0.01$) [65]. The strength of the counseling relationship impacted the magnitude of improvement.

A randomized study in preventive cardiology found that genetic counseling coupled with polygenic risk assessment improved behavioral outcomes [66•]. Patients who received a polygenic risk score and discussed it with a genetic counselor were more likely to share their risk with their family (OR, 3.31 [CI, 1.2–6]; $p = 0.02$).

While this study examined behavioral outcomes, to date, no randomized studies have examined psychological outcomes from cardiovascular genetic counseling. Surveying the broader genetic counseling literature, a systematic review noted genetic counseling has a positive impact on knowledge, perception of risk of genetic disease, disease-specific worry, intrusive thoughts, and depressive symptoms [23•]. As cardiovascular genetic counseling research matures, we will see more multicenter studies and randomized controlled trials, some of which are already under way.

Innovations to Increase Access

Both embedding genetic counselors in cardiology clinics and growth in the number of genetic counselors specializing in cardiology have increased access to cardiovascular genetics services. However, access is still significantly limited: if the incidence of inherited cardiovascular disease is expected to be 1 in 100 individuals, then the approximately 500 cardiovascular genetic counselors in the country is not enough to meet this need [67–70]. The vast majority of cardiology departments do not have a genetic counselor on staff. Most cardiovascular genetic counselors work in university medical centers [3]. In recent years, innovations intended to increase access have emerged. Some of these have been studied, though others have not.

One such innovation is genetic counseling via phone or video (telehealth). Telehealth genetic counseling reduces or eliminates the need for patients to travel to a specialized center, with the potential to increase access to patients who do not live close to one. Despite this potential, concerns about moving genetic counseling out of the clinic include losing direct communication with other members of the care team and losing non-verbal communication.

Telehealth has not been studied in cardiology specifically. However, it has been examined in oncology. A 2018 review of studies comparing telehealth with in-person services in cancer genetic counseling found that knowledge, psychological outcomes, and patient satisfaction were equivalent [71]. Findings on patient-counselor communication were mixed with one study of telephone-based genetic counseling finding patients rated communication and their own engagement lower. However, the two other studies that evaluated communication found no difference between in-person care and telehealth. A

recent survey of genetic counselors found that many who practice via telehealth are satisfied with it [72]. However, nearly all respondents noted that the biggest barrier to the broad implementation of telehealth is limitations in billing and reimbursement. This may be changing with recent shifts in national and state policies on telehealth billing [73].

In recent years, multiple private companies have started providing such services. In addition, several clinical genetic testing laboratories have started to offer telehealth genetic counseling to patients they test, often for free. These services can fill in the gaps for cardiology clinics that see patients with inherited cardiovascular disease but do not have a genetic counselor on staff or nearby.

Other innovations aim to help genetic counselors increase the number of patients they can see, thus increasing the capacity of the existing workforce. The emergence of the genetic counseling assistant role is one example of this [74, 75]. Genetic counseling assistants cover tasks that are either below or at the low end of genetic counselors' scope of practice, such as preparing test requisition forms, stocking test kits, and obtaining insurance authorization. Pirzadeh-Miller and colleagues found that adding a genetic counseling assistant to the team increased the number of patients genetic counselors could see by 60% [75]. This trend fits with the maturation of a young field. In many other areas of medicine, clinicians' roles have shifted over time to spend more time on more advanced and clinically focused tasks with other members of the care team taking over clerical work and other tasks that require less expertise. Notable examples are nursing aids, medical assistants, and speech language pathology assistants.

Increasingly, genetic counselors are employing technology to augment the care they provide patients in the hopes this will both improve outcomes and free up counselors to see more patients [76–78]. This includes the use of videos, web-portals, and chatbots to provide results and education. The field is also experimenting with triaging which patients can have their needs met entirely by such tools, so that genetic counselors can focus on higher-need patients. Biesecker and colleagues performed a randomized trial to compare a web-based platform with in-person genetic counseling for patients receiving results of genetic testing for recessive carrier status. The rationale was that these are results with lower complexity and psychological impact and thus less need for genetic counseling. The web-based platform was found to be non-inferior to in-person genetic counseling on measures of knowledge, test-specific distress, and decisional conflict [79].

One major innovation needed to improve access to genetic counselors in the USA is recognition by the Center for Medicare and Medicaid Services (CMS). As of this writing, genetic counselors cannot be reimbursed by Medicare. This requires an act of congress, which would add genetic counselors to CMS' list of recognized professionals. The National Society of Genetic Counselors has been working with members of congress, patient organizations, and other professional organizations to get this accomplished.

Insights into Inherited Cardiovascular Disease

There has been a rapid increase in knowledge about inherited cardiovascular disease, which is informing the practice of cardiovascular genetic counselors and cardiologists alike. Genetic counselors are not only guided by these studies; many of them are leading or contributing to this research as investigators. One example of insights into prognosis and risk that have come from studies led by genetic counselor-investigators is the growing body of work that demonstrates that exercise impacts both the development of arrhythmogenic right ventricular cardiomyopathy and its severity [80–82]. Within the psychological realm, recent studies led by genetic counselor-investigators have revealed the severe and traumatic emotional impact of sudden cardiac death in the family [83] as well as the challenges athletes with inherited cardiovascular disease face when their cardiologist recommends they restrict their exercise regimen [84, 85]. Genetic counselors have also led studies on genetic testing such as examinations of the yield of genetic testing [86, 87] as well as studies on genetic test interpretation [47, 53]. Research like this has helped cardiologists and genetic counselors alike better understand inherited cardiovascular disease and how to best care for patients.

Conclusions

Genetic counselors are increasingly being embedded within cardiology clinics, allowing genetic counselors and cardiologists to work closely together to care for patients with inherited cardiovascular disease. Evidence from cancer genetics and clinical psychology suggest this move toward integrated care will improve access, timeliness, and uptake of genetics services. As the field of cardiovascular genetic counseling grows, so does its evidence base. Recent studies demonstrate the value of family history assessment by a genetics specialist, careful genetic test selection, and the need for expert genetic test interpretation. Early non-randomized studies show that cardiovascular genetic counseling has a positive impact on patient knowledge, family communication, and psychological well-being. Randomized controlled studies are needed to more rigorously assess these benefits. Further work is also needed to examine which counseling approaches are most effective. As the field of cardiovascular genetic counseling continues to grow and mature, so will its scientific pursuits and evidence base.

Compliance with Ethical Standards

Conflict of Interest Tia Moscarello declares she has no conflicts of interest. Colleen Caleshu declares intellectual property and receives royalties for genome interpretation technology; she is a stockholder of Personalis, an advisor to Phosphorus and Gene Matters, and a consultant for Clear Genetics and Myokardia.

Human and Animal Rights and Informed Consent This article does not contain any studies with human or animal subjects performed by any of the authors.

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29.• Helm BM, Freeze SL, Spoonamore KG, Ware SM, Ayers MD, Kean AC. The genetic counselor in the pediatric arrhythmia clinic: review and assessment of services. J Genet Couns. 2018;27:558–64 **Helm et al. describe the establishment of a multidisciplinary pediatric arrhythmia clinic. After screening the pediatric electrophysiologists' clinic schedule, the team determined that 14.4% were appropriate for genetic counseling, most often due to a personal history of disease (75% of patients). The most common indications were long QT syndrome or other possible cardiac channelopathy**.
30.• Reuter C, Grove ME, Orland K, Spoonamore K, Caleshu C. Clinical cardiovascular genetic counselors take a leading role in team-based variant classification. J Genet Couns. 2018;27:751–60 **Reuter et al. surveyed cardiovascular genetic counselors, almost all of whom work with a cardiologist who specializes in inherited cardiovascular disease. Nearly all (95.7%) of genetic counselors surveyed gather additional data on the variants identified via genetic testing, and most (81.4%) assess pathogenicity. A role historically thought to be the purview of laboratories has been taken on by clinicians, and qualitative responses suggest this practice started due to inconsistent interpretations between laboratories, and mismanagement of families based on genetic test results**.
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