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## Scaling Genetic Counseling in the Genomics Era

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### Keywords

genomic counseling, genomic sequencing, precision medicine, genetic counseling

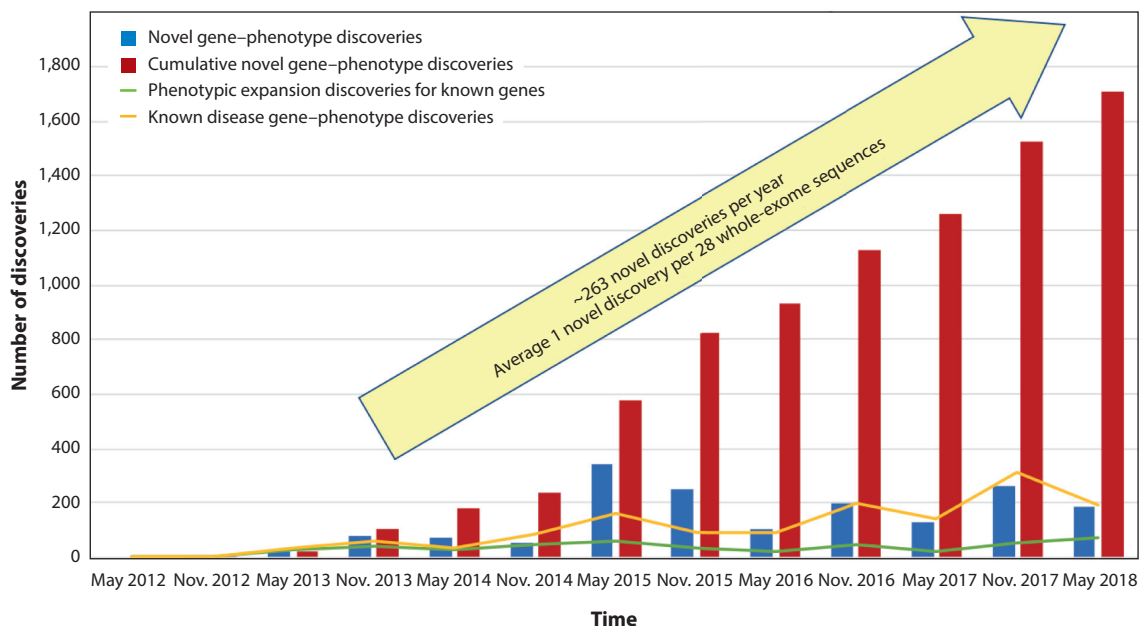
### Abstract

The development of massively parallel sequencing–based genomic sequencing tests has increased genetic test availability and access. The field and practice of genetic counseling have adapted in response to this paradigm-shifting technology and the subsequent transition to practicing genomic medicine. While the key elements defining genetic counseling remain relevant, genetic counseling service delivery models and practice settings have evolved. Genetic counselors are addressing the challenges of direct-to-consumer and consumer-driven genetic testing, and genetic counseling training programs are responding to the ongoing increased demand for genetic counseling services across a broadening range of contexts. The need to diversify both the patient and participant groups with access to genetic information, as well as the field of genetic counseling, is at the forefront of research and training program initiatives. Genetic counselors are key stakeholders in the genomics era, and their contributions are essential to effectively and equitably deliver precision medicine.

## 1. BACKGROUND

More than a decade ago, massively parallel sequencing (MPS) technology was introduced to the field of genetics, with the “potential to dramatically accelerate biological and biomedical research” (85, p. 1135). The impact of applying MPS-based exome and genome sequencing (or genomic sequencing) technology to clinical genetics was noted soon after (14). Studies exploring the transition from sequential, single-gene tests to MPS-based panel testing in the hereditary cancer setting have highlighted the time- and cost-saving benefits of multigene panels, the challenges of returning variants in genes with less well characterized disease risks, and the potential for improving diagnostic rates but also increasing the number of variants of uncertain significance identified (48, 108). Incorporating genomic sequencing into the diagnostic setting for a range of phenotypes has also led to explorations of how to maximize cost-effectiveness and clinical utility (18, 69). The utility of genomic testing for rare genetic diagnosis is established (47, 50), and new gene–disease associations discovered from MPS testing have expanded the range of conditions for which diagnostic testing is possible. In 2019, the Centers for Mendelian Genomics reported that they had identified 3,617 disease gene–phenotype pairs beyond those previously reported in association with Mendelian conditions (74) (**Figure 1**), and a 2018 report identified approximately 75,000 genetic tests currently on the market, with an average of 10 new tests added every day (71). The application of MPS technology to the generation of pharmacogenomic (44) and polygenic risk information will likely drive further growth in genetic test availability (44, 107).

Access to genetic risk information through clinically ordered and direct-to-consumer (DTC) genetic testing is increasing, driven partly by the ability of MPS-based tests to generate large



**Figure 1**

Number of gene–phenotype pairs discovered by the Centers for Mendelian Genomics over time. The blue bars represent novel gene–phenotype discoveries. The green line shows the number of phenotypic expansion discoveries (phenotypic features extending beyond those previously reported for a Mendelian condition), and the yellow line shows the number of known disease gene–phenotype pairs discovered. Figure adapted from Reference 74 with permission from the American College of Medical Genetics and Genomics.

amounts of sequencing information at little additional marginal cost. Additionally, increasing access to genomic testing, specifically in medically underserved and diverse racial/ethnic populations, is a current focus in clinical and research genetics (11, 38, 45, 95). The results of this genetic testing can be complex, and the clinical significance of identified genomic variation may be uncertain. This has become increasingly true as the range of genetic conditions that can be tested for and the types of genetic tests that can be ordered have expanded. Genetic counselors are now charged with supporting decision-making and informed consent for this broadening range of conditions and tests. The task of tracking available tests, identifying which patients are eligible for a given test, and navigating how to get these tests paid for has also become part of the scope of practice for genetic counselors in clinical and laboratory settings (13, 27, 91). Finally, genetic counselors are specifically trained to interpret, communicate, and contextualize the complex results from genetic testing while also addressing related patient and family psychological implications. Thus, the genetic counseling profession has a central role in this transformation to genomic medicine.

The first use of the term genomic counseling in relation to genetic counselors appeared in the literature in a 2010 paper (64) that suggested a critical role for genetic counselors in supporting the integration of risk information from genomic sequencing into preventative medicine. Several subsequent papers discussed the potential impact of MPS technology on the practice of genetic counseling (23, 55, 65), noting the likelihood of a shift from counseling about single-gene Mendelian conditions to complex and lower-risk diseases; an increased focus on disease prevention in addition to diagnosis; and an expanding, more directive role of genetic counselors in health promotion.

Genetic counseling has evolved, and genetic counselors have adapted in many ways in response to the paradigm shift, from practicing clinical genetics to practicing genomics. For the purposes of this review, we focus on how the practice and field of genetic counseling has changed as MPS-based genomic tests became integrated as standard of care in clinical genetics. We explore changes in genetic counseling delivery models and practice settings and discuss roles and challenges for genetic counselors related to DTC and consumer-driven genetic testing. We summarize how the field has responded to the increased demand for genetic counseling expertise and the expanding scope of practice. Finally, we discuss ongoing challenges and future directions for genetic counseling research and practice.

## 2. DEFINITIONS OF GENETIC COUNSELING

The National Society of Genetic Counselors (NSGC) defines genetic counseling as “the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease” (78, p. 77). The Accreditation Council for Genetic Counseling (ACGC), which is responsible for accrediting genetic counseling training programs, has established 22 practice-based competencies required for the successful practice of an entry-level genetic counselor across the domains of genetics expertise and analysis; interpersonal, psychosocial, and counseling skills; education; and professional development and practice (1). More recently, a need to define usual-care genetic counseling in the setting of genomic research was identified. This led to a proposed definition of genetic counseling for use in research that highlights the client-centered relationship, determination of genetic risks, tailored education, testing and management guidance, informed choice, and the promotion of adaptation and psychological well-being (15). While variations in practice exist among subspecialties and individual counseling styles, the core elements of client-centered genetic education, informed choice, and discussion of related psychological issues for the patient/participant and their family are consistently identified as key elements of the genetic counseling process.

### 3. THE EVOLUTION OF GENETIC COUNSELING PRACTICE

#### 3.1. Exploring Alternative Service Delivery Models

Traditional models of genetic counseling have commonly included a face-to-face pretest conversation between a genetic counselor and a patient that focuses on education and informed decision-making, followed by a second in-person results disclosure meeting to review results and related medical management recommendations, discuss psychological implications, and coordinate family cascade testing, as indicated (25, 103). More recently, genetic counselors have explored adapting this practice model to respond to the increase in patients who are eligible for genetic testing or have genetic test results that require genetic counseling; the expanding reasons or motivations for patients to seek genetic counseling; and, more generally, increased access to genetic counseling services (87).

One approach has been to implement delivery models that improve genetic counselor efficiency. Offering group pretest genetic counseling and providing a choice of phone results disclosure both save time for genetic counselors (24, 88). The addition of genetic counseling assistants to the medical genetics clinic team is another way to increase genetic counselors' efficiency by enabling genetic counselors to work at the top of their scope of practice and focus primarily on direct patient care (35, 72). Incorporating technology into the delivery of pre- or posttest genetic counseling has also been explored as an approach to scale genetic counseling practice. In the pretest setting, studies have developed web-based education platforms (70) and evaluated online decision aid tools (5, 16) to support informed decision-making. An exploration of patient experiences incorporating chatbot technology into consent, posttest counseling, and facilitation of family testing identified support for this model (81). The importance of ensuring that technologies that support the delivery of genetic information are accessible and engage all patients, particularly those that are underserved in genetics, has been highlighted in the literature (86).

Research on outcomes of different genetic counseling and testing models in the cancer genetic counseling setting suggests the overall acceptability of several alternative approaches (51). It is likely that more than one model will be effective and that there will be variation in efficacy and patient satisfaction within each approach. The incorporation of adapted service delivery models across the practice of genetic counseling will likely be iterative and influenced by variables at the institutional, genetic counselor, and patient levels.

#### 3.2. Incorporating Non-Genetics Providers

Unsurprisingly, genomic medicine has also made its way beyond the genetics clinic. For example, primary care physicians (PCPs) have, at times, become first-line providers identifying patients with medical and/or family histories warranting a genetic evaluation or for patients presenting with DTC genomic test reports (34). In some institutions, a genome-first approach—where genomic testing is offered to all patients through a non-genetics provider, regardless of personal or family history of disease, and genetics providers are engaged only if the findings warrant doing so—has been implemented successfully (46a, 84). Nonetheless, the arrival of genomics into the clinical practice of non-genetics medical specialists has led to increased awareness of the need for educational initiatives to prepare the non-genetics provider workforce as key stakeholders in this process (28).

Identifying this evolving area of non-genetics provider practice, recent studies have evaluated the role of PCPs and non-genetics specialty providers in genomic medicine. Challenges in provider confidence and educational needs, and varying levels of perceived utility of genomic testing, have been identified by studies exploring the involvement of PCPs in genomic test

implementation (37, 46, 54, 75). However, a small number of studies have demonstrated that, while providers had various comfort levels with genomic testing and concerns about incorporating genomic sequencing into practice, most successfully provided appropriate clinical recommendations and planned to refer to genetics specialists when necessary (22, 102). It is important to note, however, that the providers within these studies were embedded into research settings where genomics support was available.

Resources to support non-genetics providers incorporating genomics into their practice have also been developed. For example, researchers in the Clinical Genome Resource (ClinGen) program (77) have created guidance for PCP involvement in genetic testing that highlights key factors in informed consent and return of genetic test results for non-genetics providers. They have proposed tiered systems for PCPs to identify patients requiring basic versus complex genetic counseling services in order to determine when referral to a genetics specialist may be necessary (32, 66). The Clinical Sequencing Exploratory Research (CSER) consortium created a publicly available resource, “Guide to Interpreting Genomics Reports,” for non-genetics providers faced with interpreting genomic reports (29).

Finally, the enlistment of PCPs and other non-genetics specialty providers into genomic medicine has created a cohort of patients being referred to genetics clinics for posttest counseling based on testing ordered by a provider who is not a genetics specialist. In this scenario, the role of genetic counselors shifts toward handling more complex counseling cases, such as challenging psychological scenarios and complex or uncertain results, which are outside the scope of practice of non-genetics professionals. Importantly, the pretest counseling patients receive by non-genetics providers may be highly variable in content. Counseling service delivery models that do not include pretest face-to-face genetic counseling will need to be evaluated and supported to ensure appropriate informed consent is provided.

It is becoming clear that genetics professionals will need to work alongside PCPs and other specialists in a multidisciplinary way to navigate the increasing demand for genetic testing. As such, research into successfully implementing multidisciplinary workflow models and designing effective educational and support resources will need to continue, and the involvement of genetic counselors in the development and evaluation of these service delivery models will be vital.

### 3.3. Expansion of Practice Settings

The skill set of genetic counselors has been applied to an increasingly diverse range of professional roles. Genetic counselors have expanded their practice beyond the traditional setting of a genetics clinic in an academic medical center to support non-genetics providers in multidisciplinary clinics (43, 67), commercial laboratories performing genomic sequencing (52), biotechnology and pharmaceutical companies (33), and companies offering DTC genetic tests (76) or telehealth genetic services. In these roles, genetic counselors apply skills developed and relevant in the clinical practice setting, but also have the opportunity to build on and apply their knowledge base in new ways.

Genetic counselors have also taken on leadership roles in several National Institutes of Health-funded research consortia focused on studying genomic sequencing implementation. Genetic counselors have led or collaborated on local and national projects exploring topics such as participant and parent empowerment, support for non-genetics providers ordering genetic tests, and challenges in genomic results disclosure (66, 68, 88a, 106). As discussed below, this expanding employment landscape in clinical care, industry and genomic research has affected genetic counselors’ professional growth opportunities as well as the training programs that prepare them for the workforce.

### 3.4. Addressing Barriers to Genetic Counseling Access

Disparities exist in access to genomic research and genetic services across geographical settings and patient race/ethnicity and demographic characteristics (73, 89). Ways to address these disparities in genomic research have been discussed in the literature, and include designing studies that consider patient diversity as well as identifying and addressing factors that influence participation in genomic studies across diverse patient communities (26, 38). In clinical genetics practice, service models that include regional genetics support centers and the identification of financial resources and sources of support have been suggested as a means to facilitate patient access outside of urban areas and for patients with financial barriers (30, 45).

Genetic counseling–related initiatives to reduce access disparities include some of the service delivery approaches discussed above (79, 88) that reduce the need for patients to travel and attend in-person counseling visits. For example, a study investigating the efficacy of video counseling specifically targeted toward Latinx patients at risk for hereditary cancer identified overall patient satisfaction (42). Genetic counselors are also involved in studies in the National Institutes of Health–funded CSER consortium (11), which enrolls primarily patients from medically underserved and diverse racial/ethnic populations with a goal of reducing barriers (logistical, geographical, language, etc.) to receiving genomic medicine services. Adapted genetic counseling approaches in the CSER consortium include online pretest education, exploring results disclosure via non-genetics providers, implementing telephone return of results by a genetic counselor outside the participant’s health system, and disclosing negative genomic sequencing results via letter. The *All of Us* Research Program is also prioritizing enrolling participants from populations that are historically underrepresented in biomedical research (6), and genetic counselors will lead the return of medically relevant results from this program via telemedicine that will be offered in multiple languages (60). As genomic medicine moves forward, ensuring representation of and access for all patient populations will continue to be essential to the success of precision medicine initiatives.

## 4. DIRECT-TO-CONSUMER AND CONSUMER-DRIVEN GENETIC TESTING

### 4.1. Background

Founded in 2006, 23andMe became one of the first DTC genetic testing companies to enter the market, offering genetic testing services to the general public without the need for an ordering physician (8). Marketing for DTC testing began at a time of increased interest in the promise of genetics, movement toward personalized medicine, and increased awareness of genetic privacy (57). The medical genetics and public health communities quickly expressed concerns about DTC testing, including the validity of the testing, patient understanding of the limitations of DTC testing, limited informed consent, and potential privacy issues related to data sharing (9, 98, 99). Advocates for the DTC testing model emphasize the rights of consumers to the availability of genetic testing and their own raw data and argue that genetics professionals should not serve as gatekeepers to genetic testing.

### 4.2. Roles of Genetic Counselors in Direct-to-Consumer Testing

The introduction of DTC testing brought a new category of patients to the medical genetics clinic: individuals seeking genetic counseling services following DTC testing. One study that interviewed practicing genetic counselors in 2016 and 2017 demonstrated that nearly half (40.1%) of surveyed genetic counselors had seen at least one patient for the sole indication of reviewing DTC testing

results (41), an increase from the 11% reported in similar studies conducted in 2011 (17). Both the NSGC and the American College of Medical Genetics and Genomics issued position statements on the importance of the availability of genetic counselors to consumers undergoing DTC testing (9, 10, 61). However, the views of clinical genetic counselors on their preparedness for follow-up counseling of DTC testing and whether this is an appropriate use of clinical time are mixed (7, 41). Additionally, this influx of patients seeking guidance based on their DTC results places an additional burden on a workforce that is already inadequate to support current genetic counseling needs arising from genetic testing performed in a clinical setting (see Section 5.1).

The introduction of online third-party genetic interpretation tools (Promethease, Genetic Gnie, etc.) added additional complexity to the DTC arena by allowing consumers to input raw data from DTC testing to obtain an interpretation of their results. This variant interpretation is typically based on a literature review or existing databases and does not include patient-specific factors or the input of genetic professionals. Follow-up clinical laboratory testing ordered based on variants in DTC reports/raw data or third-party data interpretation reports at one commercial laboratory showed notable false positive rates and discrepancies in variant interpretation (90). Genetic counselors have also had a role in interpreting the clinical significance of these third-party interpretations. Interviews of genetic counselors exploring their experiences with online interpretation of raw DNA data described challenges inherent to these counseling sessions, such as correcting misinterpretations and patient confidence in the raw data interpretation, particularly when patient expectations do not align with the interpretations provided by the genetic counselor (7).

### 4.3. The Future of Genetic Counselors in Direct-to-Consumer and Consumer-Driven Testing

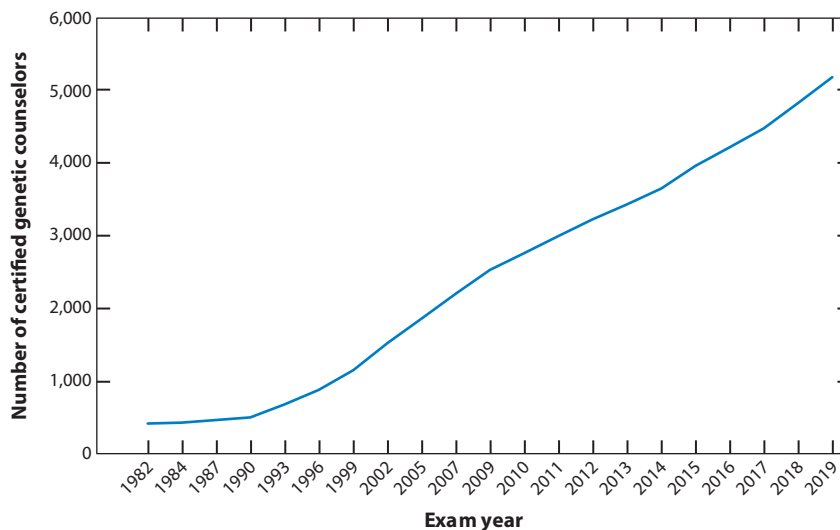
While genetic counselors have expressed discomfort with many aspects of DTC testing, they also recognize some benefits, including increased access to genetic testing, increased genetic knowledge, and provision of data for research purposes (41). To move forward, solutions must be found to combine increased testing access for the general public with increased awareness and accessibility to genetic counseling services for all consumers (53). These challenges will continue on a larger scale as DTC testing branches out to exome and genome sequencing and participants in genomic research gain access to their raw sequencing data (83). Models for roles of genetic counselors in DTC testing already exist (36). The profession will need to continue to adapt to optimize these roles, whether through the incorporation of genetic counselors into DTC services, the identification of genetic counselors specializing in follow-up of DTC results, or clinical genetic counselors expanding their clinic population to include DTC testing patients.

The evolution of DTC testing has also led to what has now been termed consumer-driven genetic testing. In consumer-driven genetic testing, consumers are not necessarily bypassing ordering providers, but are driving requests for genomic testing from PCPs, genetics specialists, and other types of specialists (76) after identifying testing options on their own. Genetics practices will need to identify innovative ways to adapt to and support a consumer-driven model, including working with the general public as well as the non-genetics providers, who may be the first to receive these requests.

## 5. SCALING THE GENETIC COUNSELING WORKFORCE

### 5.1. Increasing Genetic Counselor Demand

The growth of the genetic counseling profession has been critical to address the changes in the field of genetics and genetic counseling practice described above. According to the 2020 NSGC



**Figure 2**

Growth in the number of certified genetic counselors over time. Figure provided by the American Board of Genetic Counseling and adapted with permission.

Professional Status Survey, the size of the genetic counseling workforce doubled from 2010 to 2020 (63). **Figure 2** shows the growth in the total number of certified genetic counselors between 1982 and October 2019.

There are currently more than 5,100 genetic counselors certified by the American Board of Genetic Counseling, and it is predicted that additional growth is needed to meet the anticipated demand for genetic counseling expertise and services (93). There was a 20% increase in genetic counseling job postings between 2013 and 2016, and the US Bureau of Labor Statistics predicts a growth rate of 27% for genetic counseling positions between 2018 and 2028 (40), specifying that this growth is directly related to ongoing technological innovations in genetic testing as well as the expansion of the types of roles in which genetic counselors are employed (93).

The NSGC Genetic Counselor Workforce Working Group, assembled in 2015, reported a shortage of genetic counselors involved in direct patient care. The group proposed two primary reasons for the shortage: the increasing demand for genetic counseling brought on by the expansion of genomics into mainstream medicine, and new roles for genetic counselors in laboratories, public health, policy, and education (40). This group also predicted that the supply and demand for genetic counselors would reach equilibrium between 2024 and 2030. Thus, it is clear that ongoing work to address this shortage is necessary.

## 5.2. Implications for Genetic Counseling Training Programs

In an effort to meet this growing need for genetic counselors, established genetic counseling graduate programs have grown in size, and new programs continue to apply for accreditation. As of summer 2020, there were 52 genetic counseling graduate programs accredited by the ACGC in the United States and Canada (3). This represents a 53% increase since 2015 (when there were 34 accredited programs), compared with a 10% increase between 2005 and 2015 (80). Additionally, the number of training slots in the 2019–2020 application cycle was approximately 2.5 times greater than it was 2007 (12, 80). The predictions of continued demand for genetic counselors

described above indicate that there is a need to continue this upward trend in new genetic counselors graduating each year. An adequate rate of growth and goal for profession size have not been established and are challenging to determine due to the many variables involved, including continued changes to genetic counseling service models, evolving roles for genetic counselors, and the changing healthcare needs of the populations served (80).

Challenges with increasing the size and number of genetic counseling training programs have been identified. The number of students a genetic counseling graduate program can accommodate is limited by the number of clinical rotation sites and supervisors available. To receive accreditation, programs must demonstrate that they have adequate rotation placements and supervisors to accommodate 150% of their expected class size (2). Programs must provide the opportunity for each student to participate in a minimum of 50 cases (defined as “client encounters”), which are supervised by a certified genetic counselor. Documentation of these 50 cases, maintained by each program, serves as evidence of a student’s development of the practice-based competencies. In October of 2019, the ACGC released revised standards of accreditation with updated criteria for an approved encounter (2). These changes support greater flexibility in service model delivery, level of student involvement, and breakdown of logged cases by specialty.

Training programs continue to have a strong focus on direct patient care (2), which can be a heavy burden on sites and supervisors, especially as program sizes increase to meet the growing need for genetic counselors. A competency-based clinical training model, where requirements are tailored to each individual student, has been suggested and explored (80); however, the ACGC Clinical Training Assessment Task Force has not identified an existing tool for measuring competency in trainees, and therefore, the current approach of a standard minimum case number has been maintained (4, 80). Furthermore, activities other than direct client encounters are considered supplemental at this time. With genetic counselor roles and positions expanding well beyond the clinic, a shift in how other experiences (such as variant interpretation, test development, utilization management, sales, and leadership) are counted toward the development of practice-based competencies may require ongoing exploration.

As mentioned above, genetic counseling training programs are faced with preparing genetic counselors for a broader range of employment opportunities, beyond the historical role of providing direct patient care in a clinical genetics setting. More than 70% of counselors surveyed in a 2019 study indicated that knowledge of genomic technologies is important for successful job performance (31). Some genetic counselors have suggested that the genomic era requires robust changes to the content of genetic counseling training in order to teach new technology, as well as to prepare students for a broader variety of job settings (39). Specific changes to graduate program curricula have been proposed as necessary to promote a core skill set, including understanding genomic technology, interpreting variant data, and translating genomic test results into meaningful information for patients (39). Training programs may be challenged to keep up with the constant changes in the genetics field by continuing to add more content to their curricula, and it has been proposed that they instead focus on developing competency-based rather than knowledge-based skills. Additionally, knowledge about genetic and genomic testing technologies taught now will be out of date within a few years of graduation; therefore, focusing on student development of critical thinking, problem-solving, and navigational skills may be an appropriate approach to prepare students for a career in genetics (80).

The genetic counseling profession has a history of adapting to and growing with change, and the core genetic counseling competencies of medical genetics, education, counseling, and ethics continue to provide a foundation applicable to the wide range of roles that genetic counselors have (104). The rate of change in the profession, including new technologies, new service delivery

models, and new professional opportunities, emphasizes the importance of continuing education for genetic counselors (31).

### 5.3. Diversifying the Genetic Counseling Profession

It has been more than 40 years since the vision of diversity in genetic counseling was introduced; however, progress is still necessary in this area (56). The genetic counseling profession remains homogeneous in many aspects, especially race/ethnicity. Of the 2,571 genetic counselors who completed the 2020 NSGC Professional Status Survey, 90% identified as White, compared with 2% of respondents who identified as Black and 0.3% who identified as American Indian or Alaska Native (63). Black, Indigenous, and people of color (BIPOC) individuals currently represent approximately 14.7% of the US population and thus are severely underrepresented in the genetic counseling profession, even in comparison with the broader healthcare industry (94). Recent and ongoing efforts within the field of genetic counseling aim to provide support to genetic counselors and students of underrepresented racial/ethnic backgrounds; to provide diversity, equity, and inclusion (DEI) training and educational resources; and to amplify the voices and achievements of BIPOC genetic counselors. Of particular note are the creation of the Minority Genetic Professionals Network in 2018 and the NSGC DEI Advisory Group in 2020 (49, 62). The Minority Genetic Professionals Network provides support for genetics professionals who identify as an underrepresented racial/ethnic group and performs outreach to these groups in order to raise awareness of the importance of genetics in healthcare and genetics-related career opportunities. The NSGC DEI Advisory Group is responsible for developing and implementing the NSGC's strategies for achieving DEI within the genetic counseling profession.

An ongoing commitment and the intentional action of genetic counselors and training program faculty will be necessary to achieve significant progress toward diversity within the profession but will likely not be sufficient. Access to genetic counseling programs is limited primarily to those with the financial means to fund their genetic counseling education. Training programs funded by donor, institutional, or state and federal training support would lower the barrier for those with limited resources. Currently, scholarships for master's programs in general lag behind those for medical school and PhD programs. Investment at this level could accelerate enrollment of diverse trainees and would be a cost-effective investment in diversifying genomic medicine delivery. Finally, as in all parts of science, the pipeline needs to be expanded at earlier levels of education. Many excellent candidates remain unaware of the field of genetic counseling.

Diversifying the genetic counseling profession will take the combination and success of multiple initiatives. Genetic counselors need to increase awareness of the profession but must also take a close look at the accessibility of practices leading to program acceptance, such as shadowing and internship opportunities, the application and interview process, and scholarship opportunities (20, 21). Ongoing support for and empowerment of BIPOC genetic counselors will also be essential.

## 6. NEW DIRECTIONS AND ONGOING RESEARCH

The future of genetic counseling practice is full of opportunities and challenges. **Table 1** presents a summary of goals for the field of genetic counseling in the genomics era, along with possible strategies to achieve them.

As changes and growth in the field of genetic counseling continue, the sphere of providers and specialists that genetic counselors interact with will continue to expand. Genetic counselors will likely continue to be called upon to design effective service delivery models, workflows, and educational initiatives, and to create systems that provide patients and families with comprehensive genomic testing and counseling while optimizing the individual roles of all providers. When

**Table 1** Goals for the field and practice of genetic counseling, along with suggested strategies for achieving them

Goal	Strategy
Address the widening sphere of providers and specialists that genetic counselors interact with	Engage genetic counselors at the forefront of the design of effective service delivery models, workflows, and educational initiatives that involve non-genetics providers
Train genetic counselors with skills to support the ongoing translation of genomic technology into practice	Conduct ongoing review and, if necessary, revision to adapt ACGC standards for genetic counselor training programs
Incorporate a more personalized counseling approach to meet the individual needs of each patient	Prioritize ongoing research on how and when to tailor information for which patients Develop and evaluate necessary tools to assess patient informational needs and preferences Conduct outcomes research on adapted genetic counseling delivery frameworks
Increase the diversity of patients and participants engaged in genomic medicine	Perform community-based research to engage stakeholders from diverse racial/ethnic, geographic, and socioeconomic communities Develop and integrate innovative service delivery models to reach all communities
Diversify the genetic counseling workforce	Support genetic counselors and students of diverse racial/ethnic backgrounds Provide diversity, equity, and inclusion training and educational resources Amplify the voices and achievements of BIPOC genetic counselors
Engage genetic counselors in research	Encourage and support the research interests of current and future genetic counselors Increase funding opportunities open to genetic counselors Provide funding for positions that support effort for dedicated genetic counseling research initiatives

Abbreviations: ACGC, Accreditation Council of Genetic Counseling; BIPOC, Black, Indigenous, and people of color.

new technologies become available and the application of current technologies broadens, genetic counselors will require training to support the translation of genomic technologies into practice and will undoubtedly be called upon to do so. As such, ACGC standards will require ongoing review and, if necessary, revision to adapt to the changing professional environment.

As genomics increases patient access to and knowledge of genetic information, genetic counselors may need to consider a more personalized counseling approach to meet the individual needs of each patient. It is unlikely that a single approach will be effective for educating and supporting all patients seeking genetic counseling (105). This tailoring will build on the central tenet of “contracting” done in genetic counseling sessions, where patient and provider goals are discussed and aligned at the outset of a session. With the increasing amounts of information that will be available to patients, this process will be more important as well as more complex. Therefore, ongoing research on how and when to tailor, and for which patients, will be important. Research on the tools necessary to assess the informational needs and preferences of patients will also be required. A framework that preserves the pretest counseling and informed consent aspect of genetic counseling and facilitates patient-led tailoring of results discussions has been proposed (82). This and similar frameworks will need to be tested, and outcomes research in this area will be necessary.

The diversity of patients and participants engaged in clinical genomics will need to increase to optimize the value of genetic information and equitably deliver personalized genomic medicine.

Studies led by groups such as those involved in the CSER consortium and the *All of Us* Research Program are paving the way to achieve these goals by ensuring that recruitment efforts reach underrepresented populations. Along with the collection of invaluable genetic information, these studies also explore optimization of results delivery across diverse populations, investigate and address barriers to care, and engage community stakeholders in the process. Community-based research that engages stakeholders from diverse racial/ethnic, geographic, and socioeconomic communities will be a necessary focus of future research initiatives, as will the development of innovative service delivery models to reach all communities. Alongside these research initiatives, diversifying the genetic counseling workforce is of the utmost importance.

Finally, the continued advancement of the field of genetic counseling requires engaging genetic counselors in research, and this has been included as an initiative in the NSGC 2019–2021 strategic plan (19). While the field continues to assess and respond to fluctuations in the demand for genetic counseling services, encouragement and support for the research interests of current and future genetic counselors is necessary. In the 2020 NSGC Professional Status Survey, 2% of respondents reported having research as a part of their job title and 20% reported having research roles within their current position (63). Historically, there have been limits on research funding available to genetic counselors, given that the terminal degree for the profession is a master's. The possibility of offering PhD tracks within genetic counseling programs remains an unresolved issue for the profession (58, 101), though some institutions offer dual graduate programs in which a student earns a PhD in genetics and a master's in genetic counseling (92, 100). In 2020, the National Institutes of Health announced the availability of administrative supplements to support genomic research training experiences for genetic counselors (59), and the first National Institutes of Health R Awards specific to genetic counseling processes and practices were announced (96, 97). Increased research education and funding opportunities open to genetic counselors will aid in driving genetic counseling research forward and help to fund positions that support efforts for dedicated genetic counseling research initiatives.

## 6. CONCLUSION

The field of genetic counseling has evolved and will continue to change as it adapts to technological advances and increased patient access to genetic information. From training to practice, traditional paradigms and roles of genetic counselors are expanding, while still maintaining the core elements of client-centered genetic education, informed choice, and discussions of related psychological implications of genomic information with patients and families. Genetic counselors are key stakeholders in the field of genomic medicine and will continue to make important contributions to the development and delivery of responsive, equitable patient care.

## DISCLOSURE STATEMENT

The authors are not aware of any affiliations, memberships, funding, or financial holdings that might be perceived as affecting the objectivity of this review.

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## Errata

An online log of corrections to *Annual Review of Genomics and Human Genetics* articles may be found at <http://www.annualreviews.org/errata/genom>