Published in final edited form as:

Am J Med Genet C Semin Med Genet. 2018 March ; 178(1): 5–9. doi:10.1002/ajmg.c.31610.

Genetic counselors on the frontline of precision health

Michael J. Bamshad^{1,2,3,*}, Pilar L. Magoulas^{4,*}, and Karin M. Dent^{5,*}

¹Department of Pediatrics, University of Washington, Seattle, WA 98195, USA

²Department of Genome Sciences, University of Washington, Seattle, WA 98195, USA

³Division of Genetic Medicine, Seattle Children's Hospital, Seattle, WA 98105, USA

⁴Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX 77030, USA; Texas Children's Hospital, Houston, TX 77030, USA

⁵Department of Pediatrics, University of Utah, Salt Lake City, UT 84132, USA

Abstract

By enabling precise genetic diagnosis and treatment there is great potential for inexpensive, accurate, and widely accessible genomic information to transform healthcare and improve the general well-being of virtually every person. To maximize this potential, approaches to genetic counseling and the role of genetic counselors will need to adapt to fit changing clinical and commercial needs worldwide. This will require overcoming multiple challenges including an inadequate workforce; development and implementation of alternate models of service delivery; integration of new technologies to improve, extend, and expand services; and support for equitable education and counseling among all populations. Genetic counselors are aptly poised to take on these challenges. The result will be better informed patients and families more capable of utilizing genetic information appropriately, making autonomous decisions about their care, and modifying their approach to disease risk to actively contribute to their health. The contributors to this issue of *Seminars* discuss how key areas of genetic counseling need to evolve and how genetic counselors can play a role in shaping the future of precision health.

The framework of genetic counseling, that is the core tenets of the field, its conceptual basis, and the methodologies used, was codified nearly five decades ago (Stern, 2009). While this framework has matured and there is increasing empirical data on the value that genetic counseling adds to healthcare (Athens et al. 2017), this framework remains dynamic. This flexibility is a manifestation of multiple forces, some internal to the profession such as the varied ways in which professional roles are defined, and others external, notably the rapidly changing and ever-expanding scope of genetic, and now genomic information available driven by technological innovation (e.g., array comparative genomic hybridization, next-generation sequencing, etc.). Nevertheless, genetic counseling remains patient-focused with an emphasis on using genetic information to assess risk of disease and provide psychotherapeutic support to facilitate understanding, independent decision-making, and adaptation. In short, genetic counseling is, at its most fundamental level, technology-driven

*Correspondence should be addressed to MB (mbamshad@u.washington.edu), PM (plmagoul@texaschildrens.org) or KD (Karin.Dent@hsc.utah.edu).

and an individual-centered approach, exactly the same properties of what is now called “precision medicine.”

Precision medicine is an approach toward prevention and treatment of disease that takes into account variability in genes, environment, and lifestyle for each individual. Precision health means using similar approaches not only for disease prevention and treatment, but also to promote health and well-being. While genetic counselors originally supported families with chromosomal disorders, birth defects, and selected rare monogenic conditions, today, genetic counselors are increasingly using precise genetic information to assess risk and support decision-making about both rare and common diseases, incorporating environmental factors in risk assessments, and providing guidance to promote an individual’s health. Any one of these roles, or some combination thereof, is what genetic counselors do routinely on a day-to-day basis. Indeed, genetic counselors are often, and perhaps more frequently than any other healthcare provider, on the frontline of precision health.

In this seminars issue of the *American Journal of Medical Genetics*, we sought to survey thought leaders and experts on some of the domains of genetic counseling that are most likely to impact precision health or be impacted by precision health. The topics covered are broad: ranging from the education of genetic counselors to the impact of technology on the practice of genetic counseling. Some of the topics covered are expansive (e.g., genetic counselors as researchers) while others are narrowly focused (e.g., U.S. state laws on clinical genetic testing), but all are topics that we think are important for the collective mindset of genetic counselors worldwide to consider, if not become familiar with, in order for the profession to fully engage in and embrace the framework of precision health. Other relevant topics such as the role of genetic counseling in the prenatal setting and for common diseases, new ethical challenges in genetic counseling, and roles of genetic counselors as policy makers and family advocates are not covered due to space limitations.

Emerging from the editorial process were a number of themes shared across the contributions to this issue including: (1) the multitude of new opportunities for genetic counselors in academics, clinical care, and industry; (2) the necessity of including environmental factors in risk assessment; (3) the importance of being inclusive and engaging typically under-represented and under-served communities in research and clinical activities; (4) the need to embrace, take advantage of, and actively develop technology to extend and expand genetic counseling services; (5) the need for genetic counseling training programs to evolve, innovate, and diversify; and (6) the perspective that genomics or precision medicine should not be viewed as a threat to the genetic counseling profession but as an opportunity for counselors to exert their expertise. We couldn’t agree more. Indeed, in the absence of new approaches to service delivery, integration of technologies to expand services, an adequate workforce, and support for equitable education, and counseling among all populations, the promises of precision medicine will not be met.

Genetic counseling, like any other healthcare profession, should be an evidence-based practice. But like other fields of medicine, there exist multiple gaps in knowledge about its practice including empirical data about needs of genetic counseling clients, the process of genetic counseling, and the outcomes of genetic counseling. Precision medicine will likely

widen these gaps and more importantly, new gaps are likely to emerge as genomics becomes further integrated into clinical care. This raises important over-arching questions such as what type of research is needed and should be prioritized to support the roles of genetic counselors? What will be the impact of doing this research or not doing it? How can genetic counselors effectively expand their roles, promote inclusivity, and embrace new technologies to provide services to a rapidly expanding and increasingly diverse population of consumers? These gaps make it all the more imperative for genetic counselors to engage as researchers. Biesecker, 2018, this issue, (**Genetic counselors as social and behavioral scientists in the era of precision medicine**) lays out the contributions that genetic counselors have made to social and behavioral research, identifies some of the most pressing gaps in knowledge for genetic counselors to address, and describes the multitude of ways in which genetic counselors can contribute to translational research that may effectively educate and empower a diverse range of genetic counseling clients to use genomic information to make informed decisions and maximize their health.

One of the most active areas of investigation over the past several years has been the return of genetic test results: what results to return? how to most effectively return them? and, what is their impact on individual's health? A major challenge to implementing precision medicine is the need for efficient and cost-effective approaches for returning genetic and genomic test results, ideally ones that are scalable and can be incorporated into different models of clinical practice. Data on what types of results can be effectively returned using web-based tools can promote more efficient use of resources and enable genetic counselors to prioritize their time for interpretation and psychotherapeutic counseling (Tabor et al., 2017; Biesecker et al., 2018). Technological solutions such as these tools and the application of technology in general has the potential to transform the practice of genetic counseling and maximize health benefits for families.

How technology could improve genetic counseling services is covered in depth by Gordon et al., 2018, this issue, (**The future is now: technology's impact on the practice of genetic counseling**). They convincingly argue that technology has the potential to help genetic counselors focus on core services (e.g., psychotherapeutic counseling) and innovate new services (e.g., telemedicine), particularly for families with a higher level of need by providing more convenient, effective and efficient platforms for obtaining informed consent, educating families and providers about genetics and health-related conditions, generating differential diagnoses, estimating risk, providing decision support, documenting encounters, managing results return, and automating follow-up. Such tools should be envisioned as genetic counselor extenders and will perhaps be more readily adopted into practice should genetic counselors participate, if not lead, their development. Nevertheless, much more research is needed for the genetic counseling community, and healthcare providers in general, to understand how to best use these tools and technologies to maximize health benefits for families. Indeed, they suggest maximizing benefit will require the "nearly seamless integration across stakeholders and healthcare systems."

There is growing demand for genetic counselors in increasingly diverse clinical settings (neurology, cardiology, hematology / oncology, general medicine, etc.) and industry (e.g., commercial genetic testing laboratories) driven in large part by contemporary sequencing

technologies that have accelerated novel gene discovery for rare conditions and enabled inexpensive and efficient genome-wide diagnostic testing. While the traditional service delivery model of time and labor-intensive face-to-face encounters with a single client is still a “gold standard,” it is not a model of service delivery that can be sustained as it requires substantial personnel, resources and infrastructure. Consequently, it is not easily scalable. This demands that new models of service delivery be developed, tested and deployed. Stoll et al., 2018, this issue, (**The past, present and future of service delivery in genetic counseling: keeping up in the era of precision medicine**) briefly review traditional models of genetic counseling, forces shaping the need for new approaches to clinical care (e.g., telehealth), and summarize new models of service delivery and how they might expand and improve access to care. A sobering caveat is that many of these models have not been widely tested and have been difficult to implement and / or sustain. Importantly, they also emphasize that many of these new models utilize technology in novel ways and require new approaches to the training of genetic counseling students.

The vital topic of addressing and expanding training opportunities for genetic counselors is discussed by Riconda et al., 2018, this issue (**Genetic counselor training for the next generation: where do we go from here?**). Genetic counselor training programs face numerous challenges: how to fit the ever-expanding knowledge of genetics and the complex ways in which genetic risk factors and the environment interact; how to meet the increasing demand for genetic counselors; how to adapt and innovate training programs to ensure the competency of counselors in emerging roles in academics, clinical service and industry; as well as how to address the potential risk of insufficient clinical training sites and supervisors as increasing numbers of genetic counselors assume non-clinical roles. All of these are major and immediate challenges to graduate programs in genetic counseling. There is likely a variety of creative solutions and no single solution is going to be most effective or the best fit, or both, for every program. As a result, programs may increasingly diversify and potentially specialize in different training tracts.

Perhaps nowhere has the roles of genetic counselors been expanding as rapidly as they have in industry. McWalter et al., 2018, this issue (**Genetic counseling in industry settings: opportunities in the era of precision health**) provide a first-hand look at the varied roles for genetic counselors in industry settings where they now represent a sizeable fraction of the profession with their numbers rapidly increasing (NSGC PSS 2016). McWalter et al. provide examples of emerging roles for genetic counselors in industry settings that capitalize on their foundational skill sets and enable them to serve as bridges with other disciplines and professionals. For example, their combined expertise in clinical and human genetics enables genetic counselors to coordinate activities between: (1) consumers (e.g., families, healthcare institutions, care-providers) and commercial genetic testing laboratories to determine relevance and appropriateness of testing; (2) sequencing technicians, bioinformaticians, data analysts, and molecular geneticists to adjudicate result reports; and (3) clinicians and scientists to foster new gene discoveries, to name just a few. These roles are perhaps analogous to those played by genetic counselors in clinical settings who coordinate medical care for families among different providers, healthcare administrators, third-party payers, etc. who have a limited knowledge of genetics. Of some concern is that the skills used by

genetic counselors in these roles are not particularly unique and accordingly counselors serving in some of these roles could be replaced by other types of professionals. Moreover, there is a lot of opportunity for automation of many of these tasks—indeed artificial intelligence programs are rapidly evolving in an attempt to adjudicate genetic variants more quickly and accurately than humans. The challenge therefore may be to either adapt training programs so that genetic counselors can offer more technical depth in these new roles (e.g., an option to pursue an additional year of training in bioinformatics) or innovate these roles to take advantage of more of the unique skills of genetic counselors.

While some roles of genetic counselors in industry settings might be considered somewhat tangential to the core identity of the profession, as Baty, 2018, this issue, (**Genetic counseling: growth of the profession and the professional**) notes, it is not a dissimilar pattern of diversification observed in many health care professions with resultant advantages and disadvantages. For example, more employment opportunities are driving demand with resultant increase in salaries, stimulating development of explicit promotion and retention criteria, and subsequent higher levels of professional satisfaction. Moreover, the expansion of roles might serve to facilitate reaffirmation of core principles and identity within the profession. The core principles that make genetic counselors and their profession unique is the combination of knowledge of human genetics and psychotherapeutic counseling skills. This theme is underscored by Wicklund et al., 2018, this issue, (**Clinical genetic counselors: an asset in the era of precision medicine**) who argue that as it becomes more commonplace for individual genetic, environmental and lifestyle information to be integrated and used to predict risk of disease and promote health, it will be important that genetic counselors reaffirm their commitment to psychotherapeutic counseling and incorporate novel and perhaps somewhat unfamiliar approaches (e.g., motivational interviewing) to encourage behavior modification (Ash 2017).

Including the impact of environmental factors on individual risk assessment, particularly in evaluation of risk for common adult-onset cancers for which there is compelling data that the environment can modify risk, is especially important for certain minority populations in which environmental risk factors are more prevalent (e.g., the prevalence of obesity is higher in African Americans and Latino Americans than in European Americans (Ogden et al., 2014)). How minority populations are engaged and served by the genetic counseling profession in the era of precision medicine is discussed by Halbert and Harrison, 2018, this issue (**Genetic counseling among minority populations in the era of precision medicine**). As they note, the historical legacy of low engagement of under-represented minority populations in both genetics research and clinical genetic services must be overcome if the benefits of precision medicine are to be equitably realized for all populations. To reduce disparities in health outcomes, Halbert and Harrison advocate culturally-tailored genetic counseling (e.g., addressing varying levels of health literacy, preferred ways of receiving information, using appropriate language, etc.) and integrated approaches that consider the interaction of factors that operate at both individual and community levels. This more culturally competent counseling model should provide better information, and support, for clients to understand the implications of genetic testing in a way that also teaches them to incorporate it into their lifestyle behaviors.

Whether the experience of using strategies to deliver genetic counseling services to individuals in minority populations with cancer discussed by Halbert and Harrison can inform approaches to genetic counseling in general is unclear, but should be explored. However, doing so depends on the ability to engage under-represented populations in genomics research and will require overcoming numerous challenges. Kraft et al., 2018, this issue, (**Engaging populations underrepresented in research through novel approaches to consent**) describe ways to overcome one specific obstacle that is ensuring that informed consent processes are tailored appropriately for recruitment of under-represented minority populations. There are many novel ways to do this and they offer examples of processes such as self-administered electronic consent that employs visual icons, videos, and concise text blocks and which includes repeated exposure to key concepts, an opportunity to ask questions, and a knowledge assessment targeting common misconceptions in human subjects research that provides immediate educational feedback for questions answered incorrectly. As they wisely note, diversity in recruitment is a necessary but not sufficient step toward ensuring diversity in research participation and in turn direct translation, rather than blind extrapolation of research findings into precise genomic care.

The process of informed consent in clinical settings is governed, in part, by individual state laws regulating genetic testing. Ideally, these laws are logically consistent with one another but as Spector-Bagdady et al., 2018, this issue, (**Analysis of state laws on informed consent for clinical genetic testing in the era of genomic sequencing**) point out, this is not the case. For example, in some states, the legal definition of a genetic test is limited to variants that cause disease—an obvious ambiguity in the context of offering for return variants that are of unknown significance or even likely pathogenic. Moreover, results are often available for conditions that are not often defined as “illnesses” (e.g., complex mental health traits). Genetic counselors need to be aware of relevant state laws that might create ambiguity or confusion in the consenting process. And as Spector-Bagdady et al. note, counselors can be an important resource for regulatory agencies as they struggle to understand the complexities of medical genetics and genetic testing.

Healthy people at large represent an even greater population of potential clients in need of genetic counseling services. Demand for services has arisen in part because the lines between genetic testing in traditional clinical settings and genetic testing for non-medical results are blurring. This reflects an accelerated and explicit shift in individuals as “patients” to individuals as “consumers” of health care, and has resulted in increasingly complex decisions about testing for consumers and management options for clinicians. Ramos and Weissman, 2018, this issue, (**The dawn of consumer-directed testing**) review the history of direct to consumer genetic testing beginning in the late 1990s (e.g., GeneTree) and describe how genetic counselors need to be prepared to help consumers and clinicians navigate the wide assortment of available genetic testing options.

Finally, while the genetic counseling profession originated in the United States, the profession has expanded to nearly thirty countries—countries in South America and many in Africa and Asia being exceptions (i.e. where genetic counselors provide few, if any, services). Ormond et al., 2018, this issue (**Genetic counseling globally: where are we now?**) explore the global expansion of the genetic counseling profession by comparing

similarities and differences in training, credentialing, and scope of clinical practice across the United States, Canada, the United Kingdom, Australia, and New Zealand. The effort highlights the diversity of roles that counselors play and the similarities of what defines a genetic counselor.

The long held goals of genetic counselors to help people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease are as relevant today as they were when initially conceived. What has clearly changed and will continue to change as the era of precision medicine unfolds, is the roles that genetic counselors play and the ways in which they will achieve their goals. Instead of taking a passive role and merely adapting to the inevitable transformation in healthcare that will occur as technology, genomics, and medicine integrate, genetic counselors must challenge themselves to engage in planning, developing, and testing new approaches to education, research, and clinical care that take full advantage of precision health for families and provide guidance for other healthcare providers. This leadership will not come without hurdles, uncertainties, and even setbacks, as being at the frontline of any transformation poses both substantial risks and the potential of gratifying rewards that will ultimately benefit the families we serve. Genetic counselors are aptly poised to take on the challenges of the burgeoning era of precision health and should play a role in shaping its future, and as is often quoted, “The best way to predict the future is to create it.”

Acknowledgments

The authors are thankful to all the contributors, our colleagues who have provided comments, and the families that we have served and who have influenced our perspectives. M.B. is supported by the University of Washington Center for Mendelian Genomics (UW-CMG) funded by the National Human Genome Research Institute and the National Heart, Lung and Blood Institute grant U54HG006493. The authors have no conflicts of interest.

References

- Ash E. Motivational Interviewing in the Reciprocal Engagement Model of Genetic Counseling: a Method Overview and Case Illustration. *Journal of Genetic Counseling*. 2017; doi: 10.1007/s10897-016-0053-8
- Athens BA, Caldwell SL, Umstead KL, Connors PD, Brenna E, Biesecker BB. A systematic review of randomized controlled trials to assess outcomes of genetic counseling. *Journal of Genetic Counseling*. 2017; doi: 10.1007/s10897-017-0082-y
- Baty BJ. Genetic counseling: Growth of the profession and the professional. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Biesecker BB. Genetic counselors as social and behavioral scientists in the era of precision medicine. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Biesecker BB, Lewis KL, Umstead KL, Johnston JJ, Turbitt E, ... Biesecker LG. Web platform vs. in-person genetic counselor for return of carrier results from exome sequencing: A randomized clinical trial. *Journal of the American Medical Association Internal Medicine*. 2018; doi: 10.1001/jamainternmed.2017.8049
- Gordon E, Babu D, Laney D. The future is now: technology's impact on the practice of genetic counseling. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Halbert CH, Harrison BW. Genetic counseling among minority populations in the era of precision medicine. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)

- Kraft S, Doerr M. Engaging populations underrepresented in research through novel approaches to consent. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- McWalter K, Cho MT, Hart T, Nusbaum R, Sebold C, ... Juusola J. Genetic counseling in industry settings: Opportunities in the era of precision health. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- NSGC. 2016 Professional Status Survey. 2016. Retrieved from <https://www.nsgc.org/>
- Ogden CL, Carroll MD, Kit BK, Flegal KM. Prevalence of childhood and adult obesity in the United States, 2011–2012. *Journal of the American Medical Association*. 2014; doi: 10.1001/jama.2014.732
- Ormond K, Laurino M, Barlow-Stewart K, Wessels TM, Macaulay S, Austin J, Middleton A. Genetic counseling globally: Where are we now? *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Ramos E, Weissman S. The dawn of consumer-directed testing. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Riconda D, Grubs R, Champion M, Cragun D. Genetic counselor training for the next generation: Where do we go from here? *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Spector-Bagdady K, Prince A, Yu JH, Appelbaum PS. Analysis of state laws on informed consent for clinical genetic testing in the era of genomic sequencing. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Stern AM. A quiet revolution: the birth of the genetic counselor at Sarah Lawrence College, 1969. *J Genet Couns*. 2009; 18(1):1–11. DOI: 10.1007/s10897-008-9186-8 [PubMed: 18985445]
- Stoll K, Kubendran S, Cohen SA. The past, present and future of service delivery in genetic counseling: keeping up in the era of precision medicine. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)
- Tabor HK, Jamal SM, Yu JH, Crouch JM, Shankar AG, Dent KM, Anderson N, Miller DA, Futural BT, Bamshad MJ. My46: a web-based tool for self-guided management of genomic test results in research and clinical settings. *Genetics in Medicine*. 2017; doi: 10.1038/gim.2016.133
- Wicklund CAL, Duquette DA, Swanson AL. Clinical genetic counselors: An asset in the era of precision medicine. *American Journal of Medical Genetics Part C Seminars in Medical Genetics*. 2018; 178(this issue)

Biographies

Michael Bamshad is Professor and Chief of Genetic Medicine in the Department of Pediatrics at the University of Washington School of Medicine and Seattle Children's Hospital. He also holds the Allan and Phyllis Treuer Endowed Chair in Genetics and Development. He has studied rare genetic conditions for more than 25 years and he and his colleagues at the University of Washington were the first scientists to use exome sequencing to discover the genetic basis of a rare condition: "Exome sequencing identifies the cause of a mendelian disorder" (2010). He is a principal investigator of the University of Washington Center for Mendelian Genomics, part of a NIH program to identify the genes underlying all rare, Mendelian conditions and has developed innovative ways to share data (i.e., MyGene2) and manage return of genetic testing results (i.e., My46) from exome and whole genome sequencing in both research and clinical settings. He co-authors a popular textbook entitled *Medical Genetics*.

Pilar Magoulas is a certified genetic counselor, Chief of the Division of Genetic Counseling, and Assistant Professor in the Department of Molecular and Human Genetics at Texas Children's Hospital and Baylor College of Medicine. She currently works as a pediatric

genetic counselor at Texas Children's Hospital and also serves as the manager for the pediatric genetics clinic. Her interests include the RASopathies, specifically CFC and Noonan syndrome, expanding access to genetics and genetic counseling services through telecounseling and other service delivery models, and advocating for advancement of genetic counselors within and between different subspecialties. Pilar serves on the Board of directors for Cardio-Facio-Cutaneous (CFC) Syndrome International, the Scientific Advisory Council for the National Foundation for Ectodermal Dysplasias, and the RASopathies Network USA, and is on the Program Committee for the American College of Medical Genetics and Genomics.

Karin Dent is a certified and licensed genetic counselor at the University of Utah and an Associate Professor in the Department of Pediatrics. Karin is the Director of the University of Utah Graduate Program in Genetic Counseling and is a past president (2011) of the National Society of Genetic Counselors (NSGC). She has been active in the NSGC and other professional organizations for genetic counselors throughout her career. She currently provides clinical genetic counseling in the Medical Genetics clinics at Primary Children's Hospital and in the Motor Neuron Disease Clinic at the University of Utah Medical Center. Her research interests include the delivery of difficult news by healthcare providers and the ethical challenges surrounding informed consent and return of results in exome sequencing and whole genome sequencing research.