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PROFESSIONAL ISSUES

Introduction to the Special Issue: Public Health Genetics and Genomics

Kirsty McWalter · Amy Gaviglio

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Abstract This special issue of the *Journal of Genetic Counseling* is dedicated to public health genetics and genomics. The seventeen papers featured in this issue span such topics as genetic counselors in public health roles, newborn screening, population screening, ethics, and health beliefs and behaviors. In this introduction to the special issue, we review some history of public health genetics and genomics, present the Centers for Disease Control and Prevention's "10 Essential Public Health Services" with associated geneticsspecificrecommendations and priorities, and briefly overview how each article ties into the world of public health genetics and genomics. We hope this issue encourages genetic counselors to visualize their everexpanding and important roles in public health genetics and genomics, as well as their contributions to improving population health.

Keywords Public health genetics · Public health genomics · Public health genetic counseling · Genetic counseling

Background

Though the intersection of genetics and public health may appear to be a new concept, the Centers for Disease Control and Prevention (CDC) understood the importance of converging these two fields as far back as 1997, when they formed the Office of Public Health Genomics (OPHG). The purpose of the OPHG is to "provide timely and credible information for the effective and responsible translation of genome-based discoveries into public health & health care" (available at http://www.cdc.gov/genomics/about/index.htm). One challenge of OPHG's mission stems from the ever-widening gap between rapid scientific discoveries in genetics and genomics and the translation of those discoveries to effectively address population health. In 2011, the CDC/OPHG gathered various stakeholders together to identify priorities and recommendations for public health genetics/genomics based on the "10 Essential Public Health Services" (Fig. 1; http://www.cdc.gov/nphpsp/essentialservices.html).

The 10 Essential Public Health Services depicted in Fig. 1 illustrates general public health activities that should be addressed by communities. The report from the 2011 stakeholders' gathering (http://genomicsforum.org/files/geno_report_WEB_w_RFI_1122rev.pdf) provided genetics/genomics-specific recommendations and priorities based on stakeholder consultation and conferencing (Table 1).

A multitude of challenges lay before the field of public health genetics/genomics as we strive to address and implement the 2011 recommendations, originally slated to be worked towards by 2017. Genetic counselors have the potential to lead (and in some cases, have been leading) these efforts, even though the field may appear to be diametrically opposed to public health at times. As genetic counselors, we focus on the (often individual) genome, yet we also attempt to understand and influence the psychology of the larger population. Our subject area is tightly tied to genetic/genomic science and data collection, yet we also utilize psychosocial approaches and community health strategies. We aim to translate personalized medicine for the individual, but also strive to turn that information into measurable outcomes for the broader population. These seeming dichotomies make the challenges of public health exciting, and highlight the skills and strengths of those working in public health genetics/genomics. Individuals in public health genetics/genomics often have to think outside the box, utilize all their powers of persuasion

K. McWalter (✉)
Genetics Program, Hawaii Department of Health, 741 Sunset Avenue, Honolulu, HI 96816, USA
e-mail: Kirsty@hawaiigenetics.org

A. Gaviglio
Newborn Screening Program, Minnesota Department of Health, St. Paul, MN, USA

Fig. 1 The 10 Essential Public Health Services. This graphic was reprinted with permission of the Centers for Disease Control and Prevention, and is available on their website at: http://www.cdc.gov/nphpsp/essentialservices.html

and education, and hone their scientific expertise as well as their political spiels, in order to make progress and bridge the gaps between two seemingly very different worlds. The groundwork has been laid for us by entities such as the CDC's Office of Public Health Genomics; it is the responsibility of genetic counselors to move forward, helping to further shape and refine the world of public health genetics/genomics so we can best serve the needs of the individual and the population as a whole.

Overview

This special issue of the *Journal of Genetic Counseling* showcases a breadth of public health genetics and genomics topics, many of which tie-in nicely to the 2011 genetics/genomics-specific recommendations based on the 10 Essential Public Health Services. While public health genetics has previously been equated primarily with newborn screening, this journal issue not only contains thoughtful and enlightening discussions of this topic, but also of expanded screening, population screening, roles and achievements of genetics specialists in public health, ethical considerations, public awareness of and opinions of genetics, and public health messaging as it pertains to genetics and common complex disease.

As genetics has shifted to genomics and our ability to detect genetic variation beyond Mendelian diseases has improved, proponents of personalized medicine tout the potential benefits of knowing one's individual genome and the implications for individual health care management. At the same time, public health advocates look to the implications for larger populations which are, in fact, necessary for the

Table 1 Genetics/Genomics Applications of the 10 Essential Public Health Services

	10 Essential Public Health Services	Recommendations for Application to Genetics/Genomics
Assessment	1. Monitor health status to identify and solve community health problems	• Use databases to monitor health of the population • Study gene-environment interaction
	2. Diagnose and investigate health problems and health hazards in the community	• Utilize family history to identify at-risk individuals • Integrate electronic health records to improve coordination of care
Policy development	3. Inform, educate, and empower people about health issues	• Improve genomic literacy of the public • Develop high school curricula for genomics • Use social marketing to teach the community about genomics in understandable language
	4. Mobilize community partnerships and action to identify and solve health problems	• Engage the community
	5. Develop policies and plans that support individual and community health efforts	• Implement policies to: • Promote accessibility of genomic technology • Focus on community education and the use of family history
	6. Enforce laws and regulations that protect health and ensure safety	• Implement: • Regulatory policies and guidelines for genomic applications • Insurance coverage for high risk individuals
Assurance	7. Link people to needed personal health services and assure the provision of health care when otherwise unavailable	• Ensure accessibility to genomic applications and services
	8. Assure competent public and personal health care workforce	• Incorporate genomics into the curricula of medical schools, nursing schools, and schools of public health • Provide opportunities for continuing education around genomics
	9. Evaluate effectiveness, accessibility, and quality of personal and population-based health services.	• Evaluate genomic tests to ensure efficacy, safety, and ethicalness • Continue efforts of evaluation groups
Research		• Fund research

Table 1 (continued)

	10 Essential Public Health Services	Recommendations for Application to Genetics/Genomics
	10. Research for new insights and innovative solutions to health problems	• Focus efforts on translational research • Develop transdisciplinary research agendas • Engage community members
System management		• Coordinate all sectors of the public health system • Capitalize on the social science aspect of public health and the hard science aspect of genomics

The information in Table 1 was compiled using information in the 2011 CDC/OPHG stakeholders' gathering report (available at http://genomicsforum.org/files/geno_report_WEB_w_RFI_1122rev.pdf)

interpretation of individual genome results. While these views may seem at-odds, they are, in fact, congruent. We propose that public health genetics and clinical (individualized) genetics healthcare provision depend upon each other. They rely on each other and enhance each other's relevance. On the one hand, research and discoveries on the individual level can lead to patient advocacy, test development, and potential drug development that will impact the larger population (http://med.stanford.edu/news/all-news/2014/03/scientists-parents-join-forces-to-identify-new-genetic-disease-in-children.html) (https://beyondbatten.org/will-herndon-fund/). On the other hand, public health initiatives can lead to increased awareness of genetics services and personal genetic status, followed by lowered disease incidence on an individual and community level. One such example is the implementation of population-based carrier screening for Tay Sachs disease in the Ashkenazi Jewish community, and the subsequent 90 % reduction in the incidence of the condition by the 2000s. (http://www.genomenewsnetwork.org/articles/08_01/Tay_Sachs_gene_tests.shtml). Particularly when one reviews the 10 Essential Public Health Services and the related genetics/genomics recommendations, it is clear that clinical genetics providers and the public health genetics workforce have opportunities to work together for mutual benefit and towards common goals.

Genetic Counseling Roles in Public Health

Increasingly, there is overlap between the clinical and non-clinical roles of genetics providers. Several papers in this issue highlight the expanding roles of genetic counselors in this respect. They also reflect the 10 Essential Public Health Services of assessment, policy development, and assurance. McWalter et al. discuss the public health activities, skills, and sources of learning of genetic counselors, including those who might not identify as public health genetic counselors, in their paper, *Public Health Genetic Counselors: Activities, Skills, and Sources of Learning*. Interestingly, most of the skills reported by participants were learned on the job, suggesting opportunities for graduate training programs to increase their public health curriculum components. In *Genetic Counselors and Health Literacy: The Role of Genetic Counselors in Developing a Web-Based Resource about the Affordable Care Act*, Mann et al. describe a public health initiative tackled by a team of genetic counselors - the development of a website dedicated to the Affordable Care Act and its implications for people with rare genetic or common complex conditions. In *Creation of a National, At-Home Model for Ashkenazi Jewish Carrier Screening*, Grinzaid et al. describe the involvement of genetic counselors in the development of a population screening program for people of Ashkenazi Jewish descent. These initiatives make it clear that the separation between "traditional" (for lack of a better term) clinical genetic counselors and "non-traditional" genetic counselors is becoming more blurred and suggest that the distinctions are becoming outdated.

Particularly for genetics specialists, the concept of providing genetic counseling not only for an individual, but for a family, is a logical next step toward expanding that service more broadly to population health. Clinical genetics service provision and its associated diagnostic successes (and failures) provide an impetus for larger-scale population genetics health initiatives which, in turn, provide larger context for the potential of genetic counseling and testing. George et al., in their paper *Aligning Policy to Promote Cascade Genetic Screening for Prevention and Early Diagnosis of Heritable Diseases*, highlight this potential of genetic service provision and make a case for policy development in this important area, especially in regards to insurance coverage. Genetic counselors are a rare subset of health care providers who have experience working with extended families. We understand that genetic information has implications beyond the proband, and we are trained in and comfortable with exploring family dynamics, discussing cascade testing, and justifying genetic testing for family members. As such, we are in a unique position –with one foot firmly entrenched in the scope of traditional clinical genetics provision, but with another foot inching over to the public health realm. We work with individuals (and families) to diagnose and manage conditions that seem exquisitely unique and rare in the context of one clinic or one town, yet understand that these conditions, on a larger scale, can potentially be identified preemptively through population screening (without the agonizing diagnostic odyssey experienced by many patients and, usually, in a more cost-effective manner).

Newborn Screening

Newborn screening (NBS) has been a successful public health genetics endeavor since the 1960s, when Dr. Robert Guthrie developed a cost-effective screening test for phenylketonuria (http://genes-r-us.uthscsa.edu/resources/newborn/overview.htm). Over the last half of the century, technology has improved to the point where each state now offers a newborn screening panel of multiple conditions. Arguably, some conditions added to newborn screening panels in recent years may deviate from the original population screening criteria established by Wilson and Jungner in 1968 but, in general, this nationwide screening program is an example of a successful effort to improve population health through screening for largely genetic diseases. So successful, in fact, that the CDC named Newborn Screening one of the Ten Great Public Health Achievements of the 20th Century (http://www.cdc.gov/mmwr/preview/mmwrhtml/mm6019a5.htm). As screening technologies continue to improve, it behooves the genetics and public health communities to consider the implications of adding more conditions that further deviate from the original screening criteria and using technologies that provide information beyond what may have been anticipated. Issues of informed consent and storage and/or research use of dried newborn screening blood spots must also be considered and addressed. These are all discussions that will require input and thoughtful discussion from genetic counselors. In fact, the National Institutes of Health recognized these needs when they required ethical concerns to be addressed in their 2012 funding opportunity exploring genomic sequencing in newborn screening (http://grants.nih.gov/grants/guide/rfa-files/RFA-HD-13-010.html).

Several papers in this special issue reference the continued need for discussions surrounding NBS. Interestingly, the papers look at different stakeholders' points of view, including genetics professionals, parents, and public health organizations. Ulm et al., in their paper *Genetics professionals' opinions of genome sequencing in the newborn period*, directly address our technical capability to significantly increase the amount of genetic information obtained via NBS. Genetics professionals must put careful thought into this possibility, from both a scientific and public health perspective. Consideration should be given to the advantages and disadvantages of such an initiative, including the benefits of increasing our genetics knowledge, while balancing the desires of the public and potential negative implications of providing information with little known clinical significance. Potter et al., in *Education and parental involvement in decision-making about newborn screening: understanding goals to clarify content*, approach the topic of NBS from a different angle – that of parents. They delve into parental understanding of NBS, reasons for pursuing or declining NBS, and ways in which public health genetics/genomics education about NBS can be utilized in this context. Wintergerst et al., in *Congenital Hypothyroidism Long-Term Follow-up Project: Navigating the Rough Waters of a Multi-Center, Multi-State Public Health Project*, provide the interesting perspective of a multi-state Regional Genetics Collaborative in their attempt to partner together to further identify and define a condition identified through newborn screening. Finally, Temme et al., in *Assessment of Parental Understanding of Positive Newborn Screening Results and Carrier Status for Cystic Fibrosis with the use of a Short Educational Video*, explore the use of a video for families facing an abnormal NBS result, and the impact this has on the genetic counseling session and associated outcomes. These papers relate to the 10 Essential Public Health Services that fall under the topics involving policy development, assurance, and research.

Population Screening

Access to genetics services has consistently been a challenge, compounded by relatively few genetics-trained specialists who tend to practice in larger cities with limited resources and colleagues. Patients in rural areas and those who are under-insured or without insurance have traditionally faced barriers in accessing genetics care. Public health genetics/genomics provides potential opportunities for alternate service delivery models (e.g., video-conferencing, telemedicine), public messaging campaigns (genetic risk factors and health decisions such as smoking; genetic components of complex, common diseases), and population screening that does not necessarily require a visit with a genetics specialist. These alternate service models open avenues for education and testing, along with the potential complications that arise when service is provided on a broad scale. For example, while population screening may provide access for patients who otherwise would not have genetic testing, there remains the issue of access to genetics specialists trained to interpret those screening results and/or provide post-screening medical management and genetic counseling.

Two papers in this special issue specifically address population screening as it relates to Lynch syndrome, an inherited cancer predisposition syndrome. Cragun et al., in *Applying Public Health Screening Criteria: How Does Universal Newborn Screening Compare to Universal Tumor Screening for Lynch Syndrome in Adults with Colorectal Cancer?*, present an interesting look at how traditional criteria for population screening apply to universal tumor screening (UTS) for Lynch syndrome. They suggest that, while UTS meets many of the original population screening criteria, some criteria are not met (similar to some of the conditions more recently added to the recommended uniform newborn screening panel). This raises points of discussion regarding the continued relevance of the original screening criteria, the appropriateness of UTS and specific newborn conditions as candidates for population screening, and the need for thorough evaluation of candidate conditions and genetic tests to balance harms and benefits.

Mange et al., in *Creation of a network to promote universal screening for Lynch syndrome*, present the Lynch Syndrome Screening Network as a model of population screening for all newly diagnosed colorectal and endometrial cancers. This is a prime example of collaboration between clinical institutions and public health infrastructure. These papers hit upon the 10 Essential Public Health Services involving policy development, assurance, and systems management.

Successful implementation of newborn and population screening programs is not limited simply to providing the service itself. The ethical issues that go hand-in-hand with any population screening initiative involving genetics must also be heavily considered. Access to care, health disparities, ownership of genetic information, and the desire (or non-desire) for public policy are issues that need to be thoughtfully addressed as clinical providers and public health practitioners partner to implement these programs.

Ethics

Increasingly, the ethical, legal, and social implications (so-called ELSI) of public health genetics and genomics initiatives are gaining more attention as issues of incidental findings, genetic privacy and confidentiality, and informed consent find themselves at the forefront of the public's mind. While many of these issues are not unique to genomics, they do require special consideration in the realm of public health. The Genetics in Primary Care Institute recently mapped out ELSI considerations as they relate to genomics (Fig. 2).

As shown in Fig. 2, with the ever-expanding role of genomics, society is at a turning point. The fields of public health, genetics, genomics, and ethics are being necessarily merged. With the increasing recognition of genetics' role in public health, there is a parallel need to study ELSI issues relating

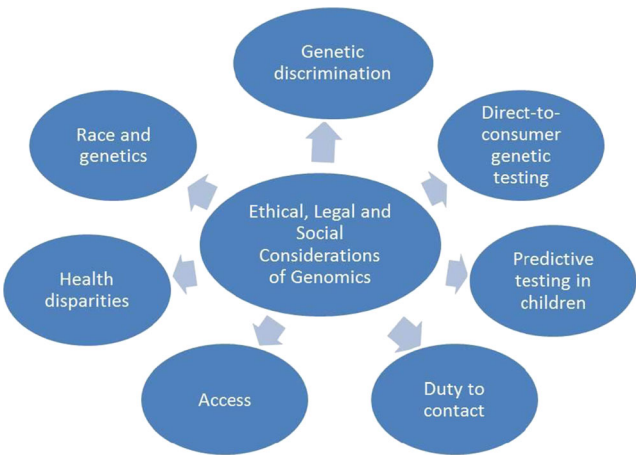

Fig. 2 ELSI Considerations Related to Genomics. This graphic was reprinted with permission of the Genetics In Primary Care Institute, and is available on their website at: http://www.geneticsinprimarycare.org/YourPractice/Pages/Ethical,-Legal-and-Social-Issues.aspx#jump-3

to genetics in the public health context. The application of ELSI into public health (PHELSI) is based on an expanded perspective, characteristic of the types of dilemmas emerging from genetics' application to public health research and practice. In many ways, the public health perspective, which applies to entire communities and populations, creates challenges calling for ethical, legal, and social approaches distinct, but certainly related, to those embraced by traditional bioethics (http://www.sph.umich.edu/genomics/issues/). The importance of ELSI in public health genetics/genomics is well-addressed by several papers in this issue. Hart et al., in *Storage and Use of Newborn Screening Blood Specimens for Research: Assessing Public Opinion in Illinois*, examine the public's opinion on a timely issue in the realm of newborn screening – the practice of storing and utilizing blood specimens from newborn screening after the testing is complete. Understanding the public's beliefs on issues such as specimen biobanking (whether newborn screening-specific, or not) and associated wide-scale genomic research is further attended to by Virani and Longstaff in *Ethical Considerations in Biobanks: How a Public Health Ethics Perspective Sheds New Light on Old Controversies*, Cohn et al. in *Increasing Participation in Genomic Research and Biobanking Through Community-Based Capacity Building,* and Martin et al. in *Perceptions of Tissue Storage in a Dementia Population Among Spouses and Offspring*. Finally, Parkman et al., in their contribution, *Public Awareness of Genetic Nondiscrimination Laws in Four States and Perceived Importance of Life Insurance Protections,* illustrate the intersection of laws governing genetics practice and the public's perceptions regarding the impact of these laws. All four of the papers, taken together, illustrate the increasing importance not only of addressing PHELSI issues, but also of the need to understand the public and engage them in these discussions. This is a component of PHELSI that will only continue to grow with the expansion of public health genetics/genomics into the realm of common, complex diseases where the development of the disease can be modified by personal beliefs and associated health behaviors. These papers hit upon the 10 Essential Public Health Services involving policy development, assurance, and assessment.

Health Beliefs/Behaviors

Often, the goal of genetic testing in common diseases is to identify individuals at increased risk for the development of a condition in order to provide preventative interventions. Towards this end, it is thought that individuals found to be at higher genetic risk may be motivated to engage in preventative behaviors, such as smoking cessation, reduced alcohol intake, or increased exercise. Understanding the intersection of genetic information with

health beliefs and behaviors and their subsequent effect on risk-reduction efforts is becoming more and more important for genetic counselors who work with individuals and populations dealing with these common conditions (i.e., all of us). Indeed, interventions and discussions around genetics and common conditions may not only need to address the associated genetic risk, but also the individual's broader attitudes towards health and perceptions of control. Because genetic explanations for a condition often become available to the public long before proven clinical utility (in part, due to the media's willingness to publicize sensationalized accounts of genetic discoveries), the relationship between the public's use of genetic explanations for disease and their associated behaviors becomes significant even before a patient reaches our office.

Two papers help lay the groundwork for a better understanding of the role of health beliefs in genetics. Thirlaway et al. look at the role of a hypothetical genetic test for lung cancer to determine the effect a positive result might have on smoking cessation. Studies such as this help to gain a better understanding of likely responses to a genetic test even before one is available clinically. Likewise, Parrott et al. explore the concepts of health beliefs and control within couples affected by a diagnosis of alpha-1-antitrypsin deficiency. This study is of particular interest in that it illustrates that, even within couples and families, differences may exist in how a disorder is perceived and further expounds on the importance of research in this area. These papers hit upon the 10 Essential Public Health Services involving assessment and research.

Conclusion

Through an examination of genetic counseling within a wide range of public health areas, we hope this issue encourages genetic counselors to visualize their ever-expanding and important roles in public health genetics and genomics. Clearly, the impact of genetics on the population's health will necessitate not only more public health genetic research, but more genetic counselors working in public health to facilitate, interpret, and translate that research. This issue of the *Journal of Genetic Counseling* only begins to touch on the wealth of new information and issues being addressed in public health genetics and genomics. We look forward to seeing genetic counselors helping lead the way to a future of improved population health.

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Human Studies and Informed Consent No research involving human participants was performed for the introduction, and so informed consent was not necessary.

Animal Studies No research involving animals was conducted by the authors.

Reference

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