A grand challenge: Providing benefits of clinical genetics to those in need

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Abstract: Genetic research, techniques, and knowledge have rapidly expanded in the last two decades with the completion of the Human Genome Project and other major advances in discovery research and diagnostic technologies. Although these developments have obvious potential, they also raise significant challenges related to programs for the actual delivery of useful genetic testing and services. This challenge is particularly acute in rural and remote areas, where lack of access to genetic services is pervasive resulting in significant inequities in access and availability of services. Huntington disease, the classic example of an adult-onset hereditary disorder, is used to illustrate this concern and highlight the imperative of exploring novel mechanisms to improve access to effective genetic services. The components of an effective and practical solution strategy are outlined, including the development of innovative delivery systems such as telemedicine, web-based education tools, and cost-reduction mechanisms. A proactive approach is essential to ensure the potential benefits, and availability of clinical genetics is realized by those in need rather than just those in reach. ***Genet Med*** **2011:13(3):197–200.**

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Genetics represents a specialized, novel, and rapidly developing field of health care. A major issue is the limited number of training programs for geneticists and genetic counselors[1] resulting in an inadequate supply of genetics professionals. For example, it is estimated that there are <3000 genetic counselors in total worldwide.[2] Compounding this issue is the paucity of access to genetic services in rural areas, as such services are usually confined to a limited number of large academic medical centers.[3] The geographic access issue in the United States is illustrated by the National Society of Genetic Counselors' survey data that reveals that there are at least 15 US states in which there are less than five reported genetic counselors in the entire state, including several, such as Wyoming and the Dakotas, in which there are one or no reported counselors.[4] Thus, for most people in rural areas, accessing genetic centers means traveling to an urban center.

The large gaps in service coverage for those outside urban centers represent a significant issue and deterrent for those who currently require genetic counseling and genetic services due to travel time, cost, and distance. As with other areas of health care, access to services is a major barrier for rural populations creating not only just a financial burden (e.g., travel, accommodation, and time off work) but also a psychological barrier in leaving family and social supports to obtain necessary care.[5] This issue is of even greater concern in terms of the future of genetic medicine as large segments of the population will be ill equipped to deal with advances in technology, treatment, and education for inherited conditions.[6] Moreover, a lack of genetic knowledge among family physicians, who are the primary, and sometimes the only, health care providers in these areas, will further compound this issue.[7]

PREDICTIVE TESTING FOR HUNTINGTON DISEASE: HIGHLIGHTING THE ISSUES

This issue is of particular relevance for a neurodegenerative disorder such as Huntington disease (HD). Predictive genetic testing for HD has been available since 1986 and represented as the first genetic test available for individuals at risk for an autosomal dominant illness.[8,9] Currently, the standard approach to such testing includes a protocol whereby at-risk individuals are required to make several visits to a genetics department to undergo genetic counseling, physical, and psychosocial evaluation.[10,11]

Uptake of predictive testing rates for at-risk individuals ranges from 5 to 25%.[12] Although such testing should be a thoughtful and individual choice, these uptake rates are low given that 65–80% of at-risk individuals suggested they would pursue such testing before the availability of the test.[13] Certainly, the absence of therapy to alter the course of illness is one component of this limited uptake.[14] However, other disincentives or barriers to testing need to be considered as they result in at-risk individuals being unable to realize the potential benefits of testing including reproductive planning, relieving uncertainty, making insurance arrangements, and getting access to services and support.[14–16] Furthermore, low-uptake rates limit the number of individuals who may become aware of research opportunities, which may lead to future treatments.

Notwithstanding personal reasons for choosing not to pursue testing, physical access to genetic services has been suggested to be a structural deterrent, which acts as a disincentive to undergoing the test.[17–19] To provide granularity to this issue, the estimated prevalence of HD is 4–8 per 100,000.[20] In the United States, this translates into a minimum estimate of 30,000 affected individuals with a further 150,000 individuals at risk for the disease.[21] According to the National Society of Genetic Counselor, only 2% of genetic counselors practice in the area of neurogenetics.[4] Therefore, even if every genetic counselor working in neurogenetics saw only those at risk for HD, this would result in 3,061 at-risk individuals for every one genetic counselor. This number, coupled with the above geographical constraints, means that under the traditional mode of genetic service delivery, individuals at risk or affected with HD may be unable to realize, or even have knowledge of, the potential benefits of future treatment, research, and psychological support.

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As novel therapies for HD become a tangible reality,[22–25] it is increasingly imperative to address and resolve this issue. Mutation-positive individuals undergo neurologic, cognitive, and structural changes in discrete regions of the brain long before the physical manifestation of the disease.[26] Thus, to be most effective in protecting the vulnerable neurons affected by HD, it is likely that therapies will need to be in place before the onset of symptoms. This will provide a strong impetus for knowing genetic risk status early, so that preventative measures may be initiated before irreversible neuronal damage occurs.

Inequalities in Access to Genetic Services

The fundamental concern illustrated by predictive testing for HD is the impact of limited access to genetic services on people at risk for HD. This issue has much broader implications for clinical genetics in general as such knowledge—when accessible—may confer multiple benefits for human health through the prevention, diagnosis, and treatment of disease.[27,28] The elucidation of genetic information will be translated into meaningful clinical benefit by influencing modes of therapy; monitoring for adverse drug reactions for those at high risk; and facilitating a profoundly different approach to disease management, especially in cancer treatment.[29,30] In addition, genetic testing will likely become routine and require frequent use by practitioners not currently using genetic medicine regularly such as family physicians and oncologists. Genetic tests will aid these physicians in practice with respect to medication prescription (to avoid adverse drug reactions), screening, and cancer treatment. Access to genetic testing and services is one of the "grand challenges" of genomics that must be addressed to ensure genetics and genomics research is beneficial for everyone.[27]

The problem of lack of access to genetic services is likely to increase as novel technologies and genetic therapies become available. Decreasing costs, such as the promise of the $1000 human genome,[31] together with rapid advancements in technologies and therapies, suggest that there is going to be an improvement in the cost-benefit impact of genetic knowledge in the health care arena.[32] However, unless there is further research to establish cost-effectiveness, as well as developing approaches to improve access, advances in genetics may lead to wider discrepancies both within and between countries.

This crisis raises significant ethical challenges. The intimate relationship between health care and health status means that barriers of access will likely result in worse health outcomes among rural populations.[33] This presents a potentially burgeoning inequity impacting an individuals' ability to function in society and live both a productive and free life.[34–38] As a result, some sectors of society will be unfairly compromised in terms of their health status in relationship to others.[39] Given that rural populations are already more likely to be uninsured, have lower incomes and less education,[40,41] and may also contain more individuals from a specific minority groups (such as Native Americans and First Nations groups), already marginalized or disadvantaged groups may suffer further disparities. Although individual autonomy (e.g., personal characteristics, preferences, and beliefs) remains a crucial component of realizing access to genetic services,[42,43] these personal characteristics are not socially determined (i.e., formed/impacted by social policy). Improving equal access to in health care involves removing these structural barriers such as physical barriers and access to services.[44]

This issue must be considered in relationship to a country's stated policy toward, and degree of commitment to, universal access to health care. For example, national health care systems such as those found in Canada and most European jurisdictions are committed to providing reasonable access to health care services across their population "without financial or other barriers."[45] In the United States, the position is more complex, with varying political and public opinions regarding universal health care and what constitutes a societal versus a personal responsibility. Nonetheless, at a minimum, there does seem to be a commitment and appeal to provide access to health care to those who desire, plan for, and request such services. President Obama's campaign is committed to "provide comprehensive and portable coverage for every American."[46] In addition, the US Department of Health and Human Services vision includes consistently leveraging genomic medicine in clinical practices across the nation and suggests information technology is key to realizing this goal.[47]

Potential Solutions

The first step in providing solutions to inequities in access to genetic services involves recognition of the problem and the realization that scientific discovery, novel treatments, and advances in genetic knowledge are the beginning, not the end of the research endeavor. To maintain momentum and funding in the genetics arena, it is imperative to realign research and financial resources to focus not just on discovery but commitment to novel mechanisms and approaches to translation. Translation in this discussion relates not just to the production of new drugs and treatment options but to actually translating research results into health care decision making and routine clinical practice. This requires a different set of research skills from creating new tests and therapies. To fully translate research from "bench to bedside" requires developing and evaluating field studies in the clinical setting by drawing on a variety of disciplines in implemental science including policy, qualitative and mixed methods studies, behavioral science, and epidemiology.[48]

Another essential component of resolving genetic access issues involves an in-depth, step-by-step, exploration of genetic counseling, testing, and service modalities to illuminate limitations and barriers. Important questions arise: what steps are needed to provide this service across geographical barriers? How can models of provision of this service (such as predictive testing) be simplified without undermining their effectiveness? What partners need to be identified to implement more widespread testing? What are innovative solutions that may be adopted by other service providers? How do we determine the need for increased capacity? And, finally, how do we mobilize appropriate resources to implement this? Specifically, in the case of HD predictive testing, these issues include knowledge of genetics and HD and availability of testing by rural health care practitioners; understanding of the benefits and limitations of testing; and the ability to assess and provide psychological and technical support. Other areas of genetic testing, such as genetic screening before medication prescription (pharmacogenomics), require a completely different approach, as these tests may not necessarily require a specialized genetics practitioner. Issues to consider in this area will include appropriate education of family physicians and rural nurse practitioners; the ability to provide testing and results quickly and clearly; and ensuring widespread uptake of testing with proven benefit. Recognizing that removing access barriers to genetic services will require multiple and different strategies, depending on the specific service to be provided.

Other areas of success in health care delivery sector broadly provide insights that can be helpful to this situation. For example, successes in immunization and awareness campaigns in the public health arena provide some insights relevant to application and expansion of clinical genetic services. These include

educating public and health care providers with novel education programs that are easily implemented, efficient, effective, and scalable. In addition, the success of vaccination campaigns involved exploring and addressing local barriers to services, such as cultural perspectives. This may be of particular relevance for communities less trusting of genetics, such as the African American population who are less likely to use genetic tests due to past discrimination.[49] Recognizing such considerations is key to developing culturally sensitive counseling and genetic testing protocols, contributing to long-term acceptability, and uptake of genetic tests.[50]

Furthermore, coordinating efforts, maintaining flexibility, and pursuing innovative methods are crucial to developing effective, efficient, and expandable strategies for equitable uptake of genetic services. Such mechanisms include the use of novel telemedicine strategies that may also allow assessment of a person's physical status remotely. This will also entail awareness campaigns to increase education among primary health care providers and the public. With telemedicine, genetic services, education, and counseling are provided remotely by telephone, videoconferencing, or using internet-based services such as Skype. Until this point, telemedicine techniques have been used in a limited capacity for clinical genetics services, primarily in the area of cancer genetics, to provide genetic counseling, test results, and follow-up care by videoconference and telephone to individuals in rural communities. Small preliminary studies have suggested that such approaches are successful, culturally acceptable, and welcome.[51,52] There still needs to be large-scale research into outcomes, together with coordinated capacity building in this area, particularly in other areas of genetics, follow-up, and assessment of patient experience and satisfaction with such services.

Another critical component to assure the successful uptake of strategies aimed at improving access to genetic services involves ensuring that approaches have support of health insurance companies (public and private) and disease advocacy groups. To reach this, data on efficacy and cost-effectiveness—taking into account both the cost of the genetic test and service provision—will be helpful. Lack of proof of the cost-effectiveness represents a key factor in the poor uptake of genotyping before warfarin prescription in the United States.[32] Moreover, research results and findings must be conveyed to policy makers, health care professionals, and the public in an accessible and direct manner by face to face meetings, marketing, and awareness campaigns. Developing strategies for translating research discoveries and pilot project findings will be essential to improving health care for the whole community. Building capacity in each of these areas will enable genetic technologies with proven utility to be implemented in a timely and effective manner and result in improved fairness and more equal distribution of genetic services for society as a whole, not just those fortunate enough to live close to a major genetic center.

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