The Need to Build Trust: A Perspective on Disparities in Genetic Testing

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IN SCREENING OR RISK ASSESSMENT TESTING, there exists the potential to improve health outcomes through early detection of genetic variation. For some conditions, such as breast cancer, Lynch syndrome, and familial hypercholesterolemia, preventive measures may be taken. The benefits of genetic testing become more apparent as evidence mounts, and at the same time we are increasingly aware that many racial and ethnic groups have limited access to these services. The resulting low participation in genetic services by minority groups poses tangible problems not only for minority individuals but also for genomic medicine as a whole, diminishing the availability of clinically useful data necessary for relevant and complete services and research to improve services.

Individuals, especially those whose family members have a genetic disease and who do not participate in genetic testing, may be disadvantaged in terms of early detection, implementing preventive actions, participating in research trials, and even accessing interventions that could slow or prevent disease progression (Glenn *et al.*, 2012). Additionally, the low participation of minority groups in genetic research and testing puts minority individuals who choose to undergo genetic testing at a disadvantage by reducing the clinical utility of the results. For example, in the *BRCA1/2* gene test, African Americans have the highest rate of receiving a variant of unknown significance (VUS), meaning that even though a mutation had been discovered, its significance could not be determined. (*BRCA* gene test is a genetic service that allows individuals to learn about their chances of developing breast and ovarian cancer. Although mutations in *BRCA1* and *BRCA2* genes are rare in the general population, mutations in these genes can raise one's chances of developing breast and ovarian cancer.) A 2006 study conducted by (Opatt *et al.* 2006) observed trends among 1765 women of various ancestries who took the *BRCA1/2* test and found that 45.1% of African American women in the study tested positive for VUS compared with only 5.5% of white women who participated in the study. Three years later in 2009, a study conducted by Myriad Genetics revealed that 16.5% of people of African descent who underwent the *BRCA1/2* genetic test had a mutation classified as VUS (National Cancer Institute, 2013). Thus, when minority groups do not participate in genetic research, effective genetic testing for people within that population is consequently diminished.

In addition to the health disadvantages encountered by groups with limited access to genetic research services, there are resulting broader consequences for the larger population. Understanding genotype–phenotype correlations across many populations furthers general understanding of health and disease and helps identify at-risk populations for certain diseases (Fine *et al.*, 2005). Disease management becomes much more difficult when researchers do not have a more comprehensive understanding of how genetic mutations affect diverse populations.

Among the reasons why minorities participate in genetic services in lower numbers than those of European ancestry are limited access to genetic services due to location and socioeconomic factors, lack of awareness of genetic services, and a distrust of how genetic information will be used. Although all of these factors are concerning and need to be addressed, the most urgent among minority groups appears to be the disconcerting lack of trust of the medical community's potential use of genetic information. The continued existence of this factor is particularly troubling because at best it should have been diminished after the passage of the Genetic Information Nondiscrimination Act (GINA) in 2008. GINA is designed to protect Americans against discrimination in health insurance and employment based on their genetic information. However, the continued low participation of minority groups over the five years since the passage of GINA suggests that there is more work to be done to combat the historical discrimination in medicine and to build trust in the biomedical research community (Roberts-Kennedy *et al.*, 2007). Members of the research community will need to engage individuals, families, and communities of diverse ancestry to better understand the core of minority concerns about genetic services in order to more effectively implement strategies that will combat these fears, build trust, and encourage greater participation in genetic testing.

Various studies assess the reasons why people of diverse ancestry take advantage of genetic testing in such small numbers. For example, a study conducted by Glenn *et al.* from 2004 to 2006 revealed that among African American, Asian, and Latina women, a leading reason why these women did not undergo a *BRCA* gene test was lack of awareness of the availability of this service (Glenn *et al.*, 2012).

Even when controlling for socioeconomic factors, a study conducted by Armstrong *et al.* revealed that African American

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women were less likely than white women to take advantage or even be aware of genetic counseling and testing services (Armstrong *et al.*, 2005). Although a different study, which was conducted by Glenn *et al.*, found that lack of awareness was a leading cause of low participation in genetic testing and counseling services among the three minority groups tested, within each group there were key differences in opinions on genetic testing. For example, the study found that among Latina women, a leading barrier to participating in genetic counseling is that it compromises traditional female roles of prioritizing the family. On the other hand, Asian women, including women of Chinese, Korean, Filipino, and South Asian origin, were more reluctant to learn about negative events in their own futures (Glenn *et al.*, 2012). Among African American women, participation in genetic testing was viewed negatively, primarily because of lack of trust in the use of their genetic information.

A caveat of these findings is that they lead to generalizations by ignoring socioeconomic, cultural, and educational diversity within each group and therefore cannot fully represent the attitudes of any one group. Nevertheless, these findings provide useful insight into the low participation in genomic medicine among minority individuals. Ultimately, identifying these issues will help clinicians better address the concerns of minorities in regard to genetic services.

The National Cancer Institute (NCI) implemented a strategy from 2000 to 2005 designed to recruit and collect data from minority groups for a cancer genetics study. When the NCI published brochures specifically geared toward these communities, minority participation increased from 1315 to 1942 individuals (Anton-Culver *et al.*, 2003).

Although distributing literature directly to minority populations had measurable results, more personalized interactions between clinicians and minority communities is another option that can yield positive results, albeit on a scale that is more difficult to measure. For example, in the late 1980s, the Mount Sinai Hospital and Northwest Baltimore neighborhood associations held a series of health fairs focused on genetic education in order to raise awareness among African American and Russian groups about the role of genetics in disease. These health fairs, however, included additional information on basic health care, such as child immunization and HIV prevention. The fairs were held in well-populated public areas, such as churches, schools, and family centers. At the end of the 30-month study period, the number of Russian individuals within the targeted neighborhoods who were screened for Tay-Sachs increased, and the number of African American people in these communities who took advantage of genetic services increased by 50%.

While distributing specialized literature to minority groups and encouraging community-based partnerships are two of the many strategies for increasing diverse participation in personalized medicine, more is needed. A U.S. Supreme Court decision made in the summer of 2013 may help to increase participation of minority individuals in *BRCA1/2* genetic testing. As many will recall, the Supreme Court ruled that Myriad's patents on these human genes were invalid,

and, as a result, other companies believe they may legally offer the *BRCA1/2* gene test. If these companies offer the test at a lower price than does Myriad, then there may be expanding numbers of women who can afford to test and consequently a decrease in the economic disparity in use of genetic services. However, without free access to a large database of variation such as the one Myriad has amassed, there is a risk that African American women and other minority groups will continue to receive a VUS diagnosis.

Even though the Supreme Court may indirectly affect the socioeconomic demographics of those who may access the *BRCA1/2* test, more direct engagement with minority individuals is important. Using a community-based partnership model, clinicians and scientists can build trust and assuage fears about genetic counseling and testing services. Until minority groups feel that they can relate to and trust members of the clinical community, they will continue to be reluctant and unwilling to take advantage of genetic services. The need to increase diversity in genomic medicine is urgent, for without recognition of the diversity of genetics, the quality and effectiveness of genetic research and services will suffer for all.

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