

MEETING REPORT

Health disparities in the genomic era: the case for diversifying ethnic representation

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Abstract

A report on the meeting 'Why we can't wait: conference to eliminate health disparities in genomic medicine', Miami, Florida, USA, 31 May to 1 June 2012.

Keywords Genomics, genomic medicine, health disparity, minority health, genetic awareness, pharmacogenomics complex diseases, community outreach, international collaboration

Cataloguing genetic differences between individuals and global populations is generating new knowledge that is having a profound impact on our understanding of biology, the practice of medicine, the development of new therapeutics and public health. The promise of genomic science cannot, however, be realized in a timely and efficient manner for all human populations unless research moves beyond predominantly individuals of European ancestry; over 90% of all genome-wide association studies so far have focused on people of European ancestry. To avoid exacerbating current inequity in health, there is an urgent need to fully engage unrepresented ethnic groups in both the discovery and the application phases of genomic science. Towards this end, this conference created a forum for diverse working groups made up of various stakeholders (including educators, public health specialists, researchers, students (medical, PhD, post-doc), doctors, researchers, genetic counselors, study coordinators, community members (for example, church liaisons), all from the United States) to (i) cultivate a shared understanding of the needs of each stakeholder to remedy health disparities in genomic medicine; (ii) identify and prioritize concerns regarding health disparities related to genomic medicine; and (iii) identify areas of maximum potential and determine projects of greatest urgency for funding.

In their keynote addresses, Donna E Shalala (University of Miami) launched the conference by stressing the importance of persistence and creativity to solve the difficult problems fueling health disparities; Vice Admiral Richard H Carmona (Canyon Ranch) emphasized the importance of genomics in medicine and highlighted family history as an important preventative tool in public health; and Eric Green, Director of the National Human Genome Research Institute (NHGRI), described the current landscape of genomics and the path towards an era of genomic medicine. He and other speakers provided several examples of how the National Institutes of Health (NIH) is supporting genomic research activities that will inform our understanding of health disparities.

Sociocultural determinants of health disparities

Olivia Carter-Pokras (University of Maryland) and several other presenters acknowledged that the major determinants of health disparities are the social, economic, cultural and political structure of societies. There was, however, recognition that most health outcomes are the result of complex interactions between factors in our environment and inherited characteristics. Thus, having access to all these types of data (socio-cultural factors, epidemiology and genetics) promises to improve understanding of how genes influence health. Chanita Hughes Halbert (Medical University of South Carolina) discussed the sociocultural differences in the use of genetic information. She pointed out that although African Americans are less likely than European Americans to use genetic counseling and testing for disease susceptibility (some of the reasons included fear of disclosure of test results to insurers, having to get the test from a specialist, race-specific marketing being negatively received, counselor's technical skills, availability of culturally sensitive counseling, genetic knowledge of diseases and unbalanced perceptions of the benefits and risks associated with knowing disease-associated genetic risks), they are, however, willing to provide samples for genetics research.

Jonathan Kahn (Hamline University) discussed the problems associated with the world's first drug directed at one ethnic group, BiDil (isosorbide dinitrate and

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hydralazine hydrochloride), approved by the US Food and Drug Administration (FDA) for the treatment of heart failure in self-identified black patients. He criticized the approval process of BiDil because it allowed the drug manufacturers to reformulate existing drugs so as to extend their patents and maintain higher prices. Furthermore, he argued that we must resist the temptation to see genomics as a neat technological fix for complex and deeply rooted social and historical problems that underlie health disparities.

Edward Ramos (NHGRI, NIH) provided data on approximately 2,000 drug metabolizing enzymes genotyped in 19 global populations from five continents. He posited that although much can be gleaned from pharmacogenomics data attributed to populations, the application of each individual's genetic variation is the ideal endpoint. His presentation provided compelling pharmacogenomic examples describing the advantages of group data as well as the implications of overgeneralization. His analysis of the global pharmacogenomic landscape led him to caution against the use of general descriptors such as 'white', 'black', 'Hispanic' and 'Asian', given the marked differences observed in allele frequencies between populations often grouped together in the same racial/ethnic construct.

How is genomics informing health disparity?

Jun Yang (St Jude Children's Research Hospital) and John Carpten (Translational Genomics Research Institute) discussed how genomics is informing cancer disparities. Yang described work on childhood acute lymphoblastic leukemia (ALL) and provided insight into the ethnic disparity in ALL survival rate in the USA, with poorer survival observed among African Americans and Hispanics than among European Americans or Asian Americans. In their published study, they observed that genomic variation that co-segregated with Native American ancestry was associated with risk of relapse in ALL. More importantly, ancestry-related differences in relapse risk were abrogated by the addition of a single extra phase of chemotherapy. Carpten showed whole-genome tumor sequencing data from black women and white women with metastatic triple negative breast cancer; he hopes that this strategy will further our understanding of the role of somatic alterations in this difficult-to-treat cancer, so reducing disparity in outcome. In collaboration with Bodour Salhia, his group has shown that although women from Egypt have high mortality rates from breast cancer, they tend to be diagnosed with luminal A breast cancer more frequently than women from other countries; this type is associated with favorable outcomes when diagnosed early. Thus, in this instance, a major cancer disparity might be eliminated through early detection and outreach.

Carlos Bustamante (Stanford University) discussed a landmark study directed at understanding the genetic basis of blond hair among Solomon Islanders. Solomon Islanders differ from the general trend of darker skin and hair pigmentation near the equator where there is higher ultraviolet radiation. Bustamante and colleagues found that an amino acid change in the *Tyrosinase-related Protein-1 (TYRP1)* gene is responsible for Melanesian blond hair, and the specific mutation underlying this trait was rare or absent outside of Oceania. He predicted that the study of more global populations will lead to the identification of many novel genetic variants with large phenotypic effects in populations currently under-represented in genomic research.

I discussed the evolution of kidney disease in populations of African ancestry, in particular missense variants in the *Apolipoprotein L1 (APOLO1)* gene that increase kidney disease risk among African Americans. I made the case for studying the parental (ancestral) populations of admixed individuals in disease mapping. Variants in the gene encoding this high density lipoprotein c (HDLc)-associated lipoprotein are present in relatively high frequency in African ancestry populations, but absent in non-African populations. These variants are thought to have increased in frequency because of a protective advantage against a deadly form of African sleeping sickness. The evolutionary understanding of the relationship between infectious disease, HDL and kidney disease is shedding light on the well documented disparities in kidney failure in African Americans compared with European Americans. This also clearly justifies studying more global populations in genomic science in the effort to understand some of the root causes of health disparities. Other examples that illustrated the need for more diversity in genomic research were presented by Esteban Burchard (University of California) for asthma, Adebowale Adeyemo (NHGRI, NIH) for diabetes and Lawrence Honig (Columbia University) for Alzheimer's disease.

Joycelyn M Lee (University of Miami) presented the work of the Genetics Awareness Project (GAP), a community education program based in South Florida designed to increase public understanding of genetics. Launched in 2009, it has already had an impact on the community, having hosted over 29 community presentations, surveys and focus groups, involving close to 1,000 participants. Preliminary data from focus groups and surveys suggest that awareness regarding genomic research and medicine are poor, and although there are a variety of misconceptions and barriers, there is a general sense of optimism and a desire to learn.

Working group summary

The working group, which included scientists, teachers students, genetic counselors and community members

among others, overwhelmingly agreed that issues related to access were most critical in reducing disparities in genomic medicine. The following access and non-access related issues were emphasized: a lack of population diversity in research, limiting its usefulness; deficits in healthcare infrastructure (for example, regarding reimbursement of healthcare costs by insurers and access to qualified professionals) and policy (for example, incentives that drive inequality, such as patents that increase the cost of care, especially medical costs); and lack of a diverse and informed general workforce. It was also emphasized that more partnerships are needed at all levels, with a strong emphasis on the need for community engagement and the attention and support of policy makers. Lastly, related to engagement, every working group touched on the importance of education of stakeholders at all levels. An informed community may be more likely to participate in research, be receptive to genomic medicine advances in care, and contribute to the diversity of the workforce. Informed physicians can ensure that advances reach all populations, particularly

those in under-served areas, and advocate for the importance of research. Informed legislators can use information in policy decisions.

In summary, although there have been some successes in which genomic medicine has met its potential and remedied a health disparity, many other early genomic medicine applications already indicate barriers to using advances. Key factors to remedy this include international genomic medicine research collaborations, dedicated research funding for the establishment of large-scale genetic epidemiology studies in underrepresented groups, enhanced community engagement, physician education and involvement, and improving access through changes in health policy and reimbursement.

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