National Human Genome Research Institute

Center for Research on Genomics and Global Health

The Role of Genomics in Health Disparities

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Abstract

In order to eliminate health disparities in the United States, more efforts are needed to address the breadth of social issues directly contributing to the healthy divide observed across racial and ethnic groups. Socioeconomic status, education, and the environment are intimately linked to health outcomes. However, with the tremendous advances in technology and increased investigation into human genetic variation, genomics is poised to play a valuable role in bolstering efforts to find new treatments and preventions for chronic conditions and diseases that disparately affect certain ethnic groups. Promising studies focused on understanding the genetic underpinnings of diseases such as type 2 diabetes or hypertension are illustrative of the positive contribution that genomics can have on improving minority health. The CRGGH has not only made significant scientific contributions with respect to these diseases but has also been involved in the development of recommendations to the NIH on the use of racial and ethnic categories; setting the agenda for genomics research in health disparity diseases; and communicating human genetic variation to the public.

Introduction

- Disparities or inequities in health refer to socio-demographic group differences in the distribution of disease, health outcomes, or access to health care.
- In the United States, there is overwhelming evidence for the existence of disparities in health when ethnic minority groups are compared to their white counterparts.
- A number of factors play a significant role in varying health outcomes, which include, but are not limited to, socio-political structure, discrimination, cultural practices (e.g., diet), socioeconomic status, exposure to harmful toxins in the environment, and access to health care.
- Genomics may inform our understanding of population differences in disease distribution and variable drug response by focusing on new insights on the global pattern of human genetic variation (HGV).

HGV: Understanding similarities and differences

Figure 1. Human migration pattern from Africa to Asia, Australia, Europe, and North and South America. Selected artifacts found around the world (1-5) are examples of evidence supporting the out-of-Africa theory.

- The frequency of genetic variations varied from region to region as a result of random chance, natural selection, and other genetic mechanisms.
- Genetic variations can occur at different frequencies in different populations, especially when those populations are widely separated and unlikely to exchange much genetic material through mating.
- There must remain a consistent link between HGV and the historic and cultural experiences of human populations as we look to understand differential disease distribution and variable drug response.

Health Disparities and Genomics

- Addressing social factors that contribute to health disparities is a priority.
- Genetics, however, can also play a significant role in understanding difference in disease susceptibility.
- There are a number of health disparity diseases that result from an interplay of social and genetic influence.

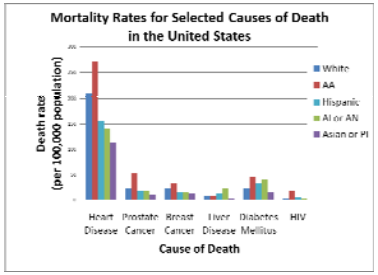

Figure 2. Death rates of selected ethnicities for six causes of death in the United States. Rates are per 100,000 population and age-adjusted to the 2000 census. AA = African American, AI = American Indian, AN = Alaska Native, PI= Pacific Islander. Source: Health, United States, 2007.

- Health disparity diseases such as type 2 diabetes is a good example of the interplay of social and genetic influence.

Type 2 diabetes GWAS in Mexican Americans

- Mano a Mano has recruited over 11,000 households (over 40,000 individuals) since 2001.
- Proposals submitted to support GWA study of type 2 diabetes on 2000 cases and 2000 controls.
- Data will complement ongoing and completed studies in China, east and west Africa, and in African American populations within the US.

Levels of causation and corresponding types of health intervention (e.g., diabetes):

I. SOCIAL STRUCTURE ← HEALTHY PUBLIC POLICY
- Race/Ethnicity
- Social Class
- Age/Gender

II. ENVIRONMENTAL INFLUENCES ← ORGANIZATION AND COMMUNITY INTERVENTIONS
- Geographic location
- Housing conditions
- Neighborhood economics
- Healthcare access, use, **clinical decision making**

III. INDIVIDUAL BEHAVIORS ← PRIMARY AND SECONDARY PREVENTION
- Smoking
- Diet
- Physical Activity

IV. PHYSIOLOGIC INFLUENCES ← SECONDARY PREVENTION
- Family history/**genetics**
- Insulin resistance
- Circulating Hormones

From McKinlay (NERI)

Conclusion

- Collaboration established with investigators from MD Anderson will allow for analysis of genetic data as well as lifestyle data such as exercise, stress, and diet.
- Understanding the detailed structure of HGV may help to deconstruct imprecise group definitions.
- Individuals cannot be treated as a representative for all those who physically resemble them or who share some of their ancestry.

"…the data will be identified as coming from one of the four populations involved, and it will be possible to make comparisons between the populations. As a result, the use of population identifiers may create risks of discrimination or stigmatization, as might occur if a higher frequency of a disease-associated variant were [associated] with a group and… overgeneralized to all or most of its members."

The International HapMap Consortium. Nature (2003).