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Genomics, Health Disparities, and Missed Opportunities for the Nation's Research Agenda

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The completion of the Human Genome Project occurred at a time of increasing public attention to health disparities. In 2004, Sankar and colleagues[1] suggested that this coincidental timing resulted in an inappropriate emphasis on the contribution of genomics to health disparities, conflating racial patterns of disease with genetic ancestry, and distracting attention from the large and compelling body of scientific evidence pointing to social determinants of health disparities.[2] For example, genomic research has emphasized discovery of genetic contributors to diabetes risk, but the recent increase in the prevalence of obesity and type 2 diabetes, which disproportionately affects minority populations, cannot be attributed to genetic changes and rather reflects social forces affecting diet, food access, and patterns in physical activity. The introduction of new genomic health technologies could also exacerbate disparities in access to high-quality health care, if specific genomic testing improved health and was only available to those who were affluent. Nonetheless, the claim persists that genomic research can reduce health disparities—if only participation by minority populations in genomic research could be increased.[3]

The source of this claim is an idiosyncratic usage of the term *health disparities* that may result in missed opportunities for the nation's health research agenda. Health disparities are generally understood to refer to systematic differences in health effects resulting from social disadvantage, but the term is often used in genomics to refer to differing health outcomes associated with population genetic variation. This usage arguably stems from the US focus

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on the association of health disparities with race/ethnicity (vs socioeconomic status), together with a growing body of knowledge about population genetic variation. Compounding the problem is a tendency in the United States to conflate health disparities and health care disparities, perhaps based on the erroneous assumption that improved health care will resolve health disparities.[4] The misunderstanding about the causes of health disparities leads to confusion about fruitful lines of research and potential remedies.

A causal association between social position and health is well established.[5] This association has been documented in both developed and developing countries and dates back to the earliest records, despite substantial change over time in the principal causes of disease. A broad array of health conditions across the lifespan follows a social gradient, wherein better health and longer lifespans track with increases in social advantage. This pattern holds whether measured by proxies of social class, such as education, income, and occupation, or by race/ethnicity.[5] In the United States, health disparities are significant and widening and have attracted considerable attention among policy makers and the general public. Yet this large body of knowledge is absent from genomics discourse, which remains largely focused on biological causes and biomedical interventions.[4]

Health care plays a crucial role in decreasing morbidity and mortality once disease processes are under way, but accounts for only a minor portion of population health status. A study comparing the major determinants of health estimated that only 10% to 15% of premature mortality could be prevented by improved or more medical care.[6] The limits of health care were demonstrated in a statistical experiment, comparing deaths potentially averted if people were to have a college education vs those potentially averted by advances in health care technology and an 8-fold difference was found favoring education.[7] Moreover, the kind of health care that makes the largest difference to population health is access to universal high-quality primary care, distinct from the specialty or high-technology care to which genomics is most likely to contribute.

Genetic susceptibility influences which individuals within a particular group experience a particular disorder. Genetics can help to explain why some African American, Native American, or Latino individuals develop diabetes, heart disease, or other common conditions whereas others living in similar environments do not, just as genetics contributes to individual variation in populations not experiencing health disparities. Research to clarify the genetic contributors to disease etiology has many potential benefits. It may help to elucidate disease mechanisms and could inform genetic tests and drug development. Inclusion of diverse populations in genetic studies will enable identification of a fuller range of genetic variation contributing to various health outcomes, potentially leading to improved genetic tests that are applicable to all populations. Well-designed gene-environment studies across multiple populations may also help to delineate important environmental modifiers of disease. All of these considerations point to the potential health value of genetic research. However, these efforts will not provide strategies for addressing the more substantial contributors to health that are rooted in social, material, and environmental conditions.

Given population genetic variation, it is to be expected that genetic effects will sometimes augment, and at other times run counter to, the effects of social disadvantage. For example,

African Americans with chronic kidney disease on average progress more rapidly to end-stage renal disease than European Americans with chronic kidney disease. Two variants in the *APOL1* gene, seen in people of sub-Saharan African descent, contribute to this disease progression.[8] Nevertheless, a comparison of African Americans with and without *APOL1* risk genotypes with European Americans demonstrated that the *APOL1*-associated risk, although significant, accounted for only about 10% of excess incidence of albuminuria (a marker of risk for end-stage renal disease) among African Americans; a disparity remains between African Americans with low-risk genotypes and European Americans.[8] In this case, the higher risk of albuminuria appears to be due to a combination of social and genetic determinants. Conversely, the incidence of acute lymphoblastic leukemia in African American children is less than half that of European American children, due in part to a difference in the prevalence of 2 risk variants. Yet survival from this malignancy is lower in African American children. This survival disparity is eliminated in a clinical setting characterized by high-quality care, aggressive case management, and financial support that eliminates out-of-pocket costs.[9] In this case, the genetic determinants lead to lower disease risk, but social determinants lead to worse outcomes for those African American children who develop the disease. These examples support the value of understanding the health implications of population genetic variation—but also illustrate that social determinants consistently reduce health outcomes in disadvantaged populations, independent of genetic risk.

Characterizing health disparities as a challenge for genomics, rather than as a challenge for health and social sciences more generally, generates several problems. It justifies studies that focus on genetic causes of complex diseases with the goal of developing medical interventions, rather than studies that assess genomics within the context of social and environmental contributors to disease. Genomics research could make a positive contribution to the elucidation of causal mechanisms of health disparities and development of potential remedies, but that dividend is likely only if such contributions are integrated into, rather than emphasized over, broader social models of disease and interdisciplinary research methods. Viewing health disparities as addressable by medical care also focuses translational science on health care innovation rather than on community-based health promotion and intersectoral policy approaches. In so doing, attention and resources are diverted away from approaches that are more likely to reduce health disparities, such as efforts to increase education levels, reduce income inequality, promote community-based dietary and exercise initiatives, and ensure universal access to primary care.

Importantly, an approach primarily based on the development of innovative health care also threatens to sideline genomic research that might offer more substantive benefit for populations experiencing health disparities. An emerging body of preliminary data related to epigenetics, the microbiome, and genetic modifiers of response to the environment points to a range of opportunities for genomic tools to elucidate the causal pathways of health disparities. Promising findings on epigenetic changes related to childhood adversity, for example, point to ways in which genomics could contribute to a better understanding of how social disadvantage is embodied and expressed. Research efforts of this kind might ultimately help policy makers to weigh priorities when allocating resources to address social determinants of health.

Health disparities are complex and multifactorial. Reducing health gaps in the United States will require researchers and clinicians from many disciplines who share a common understanding of key terms and the role of social determinants of health to work in close collaboration with affected communities. Resolving a fundamental misunderstanding about the relationship between genomics and health disparities may create new opportunities for research collaborations, allowing large research investments in genomics to be leveraged for promising population health research. Failure to do so has the potential to deepen mistrust of scientific research and health care among those populations most burdened by health disparities.

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