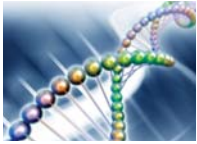


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The role of genetics in understanding racial and ethnic health disparities

The use of race and ethnicity as surrogate markers for describing risk for disease on a genomic level is becoming increasingly common in both clinical practice and research settings.^{1,2} The utility of race to predict ancestry, genetics, and outcomes of treatment of a patient has been described as the *practitioner's dilemma*.³ Both race and ethnicity carry complex connotations that reflect culture, history, and socioeconomic and political status, as well as an important connection to genetic ancestral origins.⁴

As our knowledge of human genetic variation is growing, data refute the notion that “races” are genetically distinct population groups and demonstrate that no sharp genetic boundaries can be drawn between human population groups.^{5,6} Self-identified race and ethnicity correlate with genetic variation between population groups, but they do not necessarily correlate with an individual patient's distinct genetic background. Thus, there is a debate and some confusion, particularly in the media, as to the relationships between genetic risk of diseases and race.

REPORTING—LOST IN TRANSLATION

We are unraveling the genetic components of common diseases where racial and ethnic health disparities exist. However, the complexity of the research findings is too often lost in translation when presented in the media. Frequently, headlines seem to send a

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message to health professionals and the public that the identification of a gene variant involved in a common complex disease is the “golden ring” for understanding the cause of a health disparity.

A *New York Times* headline states, “Study Points to Genetics in Disparities in Preterm Births.” The accompanying image is a poignant photograph of a premature black baby.⁷ The article describes the significantly higher rates of premature births among black women compared to white women. This headline draws directly from the

media collaborate to accurately report research findings.

THE COMPLEXITY OF RACIAL- AND ETHNIC-SPECIFIC GENETIC SCREENING GUIDELINES

One area where the concepts of race, ethnicity, and ancestry are often inflated is recommendations for screening for genetic conditions such as sickle cell disease, alpha-thalassemia, beta-thalassemia, and cystic fibrosis. Current guidelines from the American College

“The role of genetics in racial and ethnic health disparities must be reported factually and responsibly by members of the media.”

researchers' conclusions but was not based on any genomic research;^{8,9} the headline itself, however, raises a provocative question of the genetic basis of the health disparity. The essential point of the research article that the headline fails to capture is that this is a cohort study of state birth and death records and not a genetic study. We need additional studies that investigate the genomic, behavioral, social, and environment determinants of health as they relate to these documented disparities.

Does this type of media reporting, by way of sensational headlines and photographs, influence health care professionals and the public to overstate the genetic contribution to differences in health and disease? The debates over genetics and health disparities are complex, and tension increases when social, political, racial, and ethnic labels are added. It is imperative that scientists and the

of Obstetricians and Gynecologists (ACOG) recommend offering carrier screening for sickle cell disease and thalassemias to persons of African, Southeast Asian, and Mediterranean descent. The guidelines note “that ethnicity is not always a good predictor of risk because individuals from at-risk groups may marry outside their ethnic group.”¹⁰ ACOG recommendations for cystic fibrosis carrier screening provide that screening should be offered when both partners are of Caucasian, European, or Ashkenazi Jewish ethnicity. The guidelines further note that it is reasonable to offer cystic fibrosis carrier screening to all couples regardless of race or ethnicity as an alternative to selective screening.¹¹

How do health professionals understand these concepts to describe groups or populations at risk and then apply them into clinical practice? There is a great need for multidisciplinary research collaborations that appro-

GENOMICS IN PA PRACTICE

“As long as social inequities and cultural differences exist, race is likely to matter in health care for a very long time.”

propriately explore the dissemination of research findings to health professionals and the general public and how they understand the constructs of race and ethnicity.¹²

►BETTER UNDERSTANDING NEEDED

The study of genetic variation is important in unraveling all the causes of health disparities.¹³ The role of genetics in racial and ethnic health disparities, however, must be reported factually and responsibly. Only with a better understanding of all the determinants of health—including genetic and social determinants such as culture, education, occupation, household income, and access to health care—combined with an understanding of the patient’s natural and built environments and how this information interacts with a patient’s genome, will

we truly have personalized medicine. Clinicians must be engaged in the realization of the benefits of personalized genomic medicine for all patients.

“Will a patient’s race have more or less clinical utility in the genome era? As our knowledge grows, race may have less relevance in decisions that are based primarily on biochemical or physiologic processes, which may be better guided by genomic information. However, as long as social inequities and cultural differences exist, race is likely to matter in health care for a very long time.”¹⁴ [JAAPA](#)

The content is solely the responsibility of the author and does not represent any position or policy of the National Human Genome Research Institute, National Institutes of Health, or Department of Health and Human Services.

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