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Genes, Race and Health Disparities

The relationship between genes, race and health produces an important debate in the genomics era. Statistics on chronic disease and mortality in America show significant health disparities between self-identified racial groups. Many people in the scientific community believe that using genomics to come up with biological components of race and applying them towards racially based treatments and therapies may be the best step toward ameliorating some of these problems. However, it is important to look critically at how race is being defined, as well as to take into consideration the many significant confounding factors that may contribute to racial disparities in health. This would allow for the implementation of appropriate and effective medical treatment and prevention plans for every patient. As the scientific community continues to move forward in its attempts to address large health disparities, it is important to take on a multi-disciplinary approach that capitalizes on the wonderful, novel genomic technology that has become available, while remaining sensitive to the bigger social contexts of disease and health.

In examining the discourse on the interconnectedness of race, genetics and health, it is important to first understand how science is attempting to define race using a biological basis. In a recent study entitled *The Genetic Structure of Human Populations*, researchers analyzed genotypes at 377 autosomal microsatellite loci (short sequences of nucleotides on the DNA) from 1056 individuals from 52 different populations around the

globe (Rosenberg et. al 2381-2385). The study was successful in identifying genetic markers that could distinguish continental ancestry among global population groups. In some analyses, these genetically based groups “resemble groups that are historically categorized as ‘races’” (Barr). Other population genetics studies have also found genetic clusters and rare genetic variants that show evidence of race specificity (Fine 2125-2128). These findings have lead many scientists and medical professionals to assert that the self reported race of a patient can be used as a proxy for risk of certain genetic characteristics, which can then be applied to predictions of medical treatment outcomes for that patient. The practice of race based medicine is supported even further by data about certain Mendelian diseases that have very apparent heightened prevalence of specific, disease causing mutant alleles in certain populations. Examples include extremely high prevalence of Sickle Cell Disease among Africans and African Americans, and Tay-Sachs Disease among Ashkenazi Jews (Fine 2125-2128).

Opponents of a genetic definition of race and “race based medicine” argue that while the Rosenberg et al study was able to categorize people using genetic markers into groups by continental ancestry, it is important to note that in the results of the study, there is substantially more genetic variation *within* each group than there is *between* them. The study showed that 95% of all human genes are identical. Of the 5% variation that remained, 95% of the variation between individuals was within a “racial” population, and only about 5 % of variation represented differences among the major groups. This means that only .25% of total genetic variation was among the ancestry groups determined by Rosenberg et al’s data analysis (The New Genomics of Race as They Pertain to Health). This data contests the argument that race should be a key component in medical

assessments because it means that many of the genetic differences that could lead to varying health outcomes are within a single racial category.

One study that exemplifies the major differences that can occur within a socially constructed racial group is a study done on disparities in infant mortality among White and Black patients in Illinois. In the study, White mothers averaged about 5.6 infant deaths per 1000 live births, while Black mothers averaged about 16.3 (The New Genomics of Race as They Pertain to Health). To study the disparity, researchers divided mothers into 3 groups: White women born in the United States, Black women born in the United States and Black women born in Africa. Results of the study showed that babies born of US born White women and African born Black women had similar birth weights, while babies born of US born Black women were more likely to be underweight (The New Genomics of Race as They Pertain to Health). This is significant because low birth weight is the primary means of infant mortality, and these results show that race alone does not provide sufficient information to account for the health disparity. Thus, basing medical treatment and prevention solely on correlations between genetics and continental ancestry would be an overgeneralization which could potentially lead to ineffective health care delivery and adverse implications on the health of individuals.

Another fear that some scientists and physicians have about using race as a proxy for genetic health risks is the failure to consider other significant confounding factors that undoubtedly contribute to racial health disparities. These include, but are not limited to, environment, socioeconomic status, stress, unconscious physician racial biases and differential access to care (Collins). There are several examples of how seemingly genetically or racially based health disparities can be explained, or at least more fully

understood, by one or more of these alternative factors. For example, studies have shown that while White women have a significantly higher incidence of breast cancer than Black women, Black women have significantly higher rates of mortality from breast cancer than White women. Meanwhile, other racial groups that have lower incidence also have lower mortality (Breast Cancer: the Molecular, and the Human). While some may be quick to attribute this disparity to a genetic mutation that somehow makes Black women more vulnerable to the effects of the cancer, or certain treatment options, others argue that it could be caused by more social discrepancies such as later stage diagnoses for Black patients, less access to preventative health care, and lower education statuses. According to one study done in Harlem, gaps in mortality rates were narrowed significantly by introducing community outreach programs that helped identify high risk women in the community, bringing them into clinics earlier, and navigating them through the health care, diagnosis and treatment process. These results alludes to the assertion of differential access and later stage diagnosis as primary reasons for disparities in mortality (Breast Cancer: the Molecular, and the Human).

Alternate explanations of health disparities by race become especially important when physicians and doctors begin using race as a basis for certain drugs or treatment plans. Supporters of genomics and race based medicine argue that genomic research supports and will continue to support concrete genetic variations such as varying distributions of polymorphisms in drug receptors or drug metabolizing enzymes between race, that legitimizes the need to develop race specific drugs therapies (The New Genomics of Race as They Pertain to Health). Many current physicians stick by their racially biased medical decisions. For example, psychiatrist Dr. Sally Satel writes in a

news paper piece, “When it comes to practicing medicine, stereotyping often works... When I prescribe Prozac to a patient who is African-American, I start at a lower dose...I do this in part because clinical experience and pharmacological research show that blacks metabolize antidepressants more slowly than Caucasians and Asians.” (The New Genomics of Race as They Pertain to Health) While some may argue that Dr. Satel’s biased approach to treatment is good in that it is best for the patient, others claim that it projects an unnecessary and possibly inaccurate stereotype onto a patient that could result in adverse health outcomes.

A great example of the debate over race based therapies and the importance of non-genetic factors comes from recent research done on cardiovascular disease in Black and White patients. One study was on the effects of a drug called Enalapril, which was tested on Black and White patients with severe Congestive Heart Failure (CHF). Results of the study showed that White patients that received Enalapril were hospitalized for a shorter amount of time than White patients that received a placebo, while Black patients saw no difference in length of hospitalization between Enalapril treatment and the placebo (Barr). Many took these results as significant evidence of genetic determinants that White patients had a more favorable drug response to Enalapril than Black patients, and that prescribed treatments for CHF patients needed to be racially tailored. However, a second study was then completed in which Enalapril was found to be equally effective for both Black and White patients exhibiting early signs of CHF in lowering severity of heart failure, preventing complete failure and decreasing mortality. Although in general, Blacks have higher absolute risk for CHF than whites, Enalapril did end up being effective for both racial groups (Barr). This is yet another example where one group of

people could have been wrongly denied a potentially successful treatment option based on racial generalizations.

Another example of race specific treatment for cardiovascular disease is BiDil, a combination of two pre-existing drugs that was actually patented to specifically treat Black patients with cardiovascular disease. BiDil works in preventing CHF by “increasing the level of nitric oxide and decreasing oxidant stress in the vascular endothelium and thereby increasing vasodilation” (Barr) The pharmaceutical company that pursued the patent for this race specific drug claimed that its proven effectiveness in Black patients was probably due to a heightened prevalence of a certain genetic mutation among Black people that reduces nitric oxide levels in the endothelial cells (Barr). However, many examiners and critics of race based treatment have pointed to alternate explanations. First, clinical trials done for BiDil are thought to be faulty in that after seeing an original data analysis that BiDil’s effects were more pronounced in Black patients than White patients, the follow up, placebo-controlled study included no White subjects (The New Genomics of Race as They Pertain to Health). Also, these kinds of testing procedures and pushing of racially specific results are thought to be a result of the corporate interests of the pharmaceutical company in getting a new drug patent (The New Genomics of Race as They Pertain to Health). Finally, and perhaps the most significant, is that the noted racial differences in vascular nitric oxide activity could stem from the fact that decreased endothelial levels of nitric oxide is a major side-effect of diabetes (Barr). Being that socioeconomic disparities have led to a significantly high prevalence of diabetes in the Black population, the pronounced effects of BiDil could have nothing to do with race at all, but rather with the presence of comorbid diabetes. Thus, approving

the drug as a race specific treatment for Black patients could be wrongly denying a potentially beneficial treatment for people of all races with CHF and comorbid diabetes.

These kinds of studies, and many others like it, emphasize the importance of careful, full spectrum research when it comes to addressing health disparities. The field of genomics plays an important role in continuing the research on genetic determinants of disease susceptibility and treatment outcomes. However, its ability to do so may be limited to examining genetic determinants of disease on an individual level, rather than trying to generalize findings to larger, racially based populations. Francis Collins, director of the National Human Genome Research Institute, writes that

A true understanding of disease risk requires a thorough examination of root causes. 'Race' and 'ethnicity' are poorly defined terms that serve as flawed surrogates for multiple environmental and genetic factors in disease causation...Research must move beyond these weak and imperfect proxy relationships to define the more proximate factors that influence health (Collins).

In order to achieve the kind of understanding Collins says we should strive for, scholars and health professionals from all different fields need to come together and tackle the problem of health disparities from all angles. Only by moving past overgeneralizations caused by race based medicine and continuing in vigorous genomic, social and community based research can progress be made toward understanding and resolving serious public health issues.

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