Race & Genes

By Ruth Hubbard
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It is beyond comprehension, in this century which has witnessed holocausts of ethnic, racial, and religious extermination in many parts of our planet, perpetrated by peoples of widely different cultural and political affiliations and beliefs, that educated persons—scholars and popularizers alike—can come forward to argue, as though in complete innocence and ignorance of our recent history, that nothing could be more interesting and worthwhile than to sort out the "racial" or "ethnic" components of our thoroughly mongrelized species so as to ascertain the root identity of each and everyone of us. And where to look for that identity if not in our genes?

Every decade or so, a new prophet arises who promises to decode the currently advertised Rosetta Stone that will enable each of us to not only know who we truly are, but to use that knowledge for our individual and collective benefit. And, of course, no period has inspired more hope and determination than the present, now that large sums of money (which many argued might be better used to serve urgent medical and other social needs) have gone into spelling out—"sequencing"—the molecular composition of the "genes" which are part of that long string of "bases" (the A's, G's, C's, and T's) curled up within the nucleus of each of the cells in our bodies. To make good on this effort has been rendered all the more urgent by the fact that some of the scientists pioneering the task have suggested that the very volume and complexity of the new information make it virtually impossible to interpret. (As one of these pioneers artfully stated: "We don't know shit" about it.) So, what to do?

Why not use the information to tackle the meaning of "race," the question that has bedevilled Europeans and both European and African Americans for a very long time? And better yet, why not use it to tackle the relationship of race to health, a question that haunts us in more recent times?

After all, we know for a fact that here in the United States, overall and by a panoply of criteria—morbidity, mortality, life expectancy, and so on—the health of African Americans is not only

different from that of European Americans, but worse. So, let us disregard such epiphenomena as racial discrimination and the associated differences in income, family structure, education, rates of employment and incarceration, and such. Let's get down to the basics—our essential nature: our DNA (or "genes"). If we could establish "racial" gene clusters and if we could use these to predict disease, and so prevent it, wouldn't we all be better off?

Several things are wrong with this plan. For one thing, the manifestations of inherited conditions can vary considerably and unpredictably from one person to another and, indeed, in the same person at different times. This is so because many factors, both within and outside ourselves, affect the ways we develop and function. This is true of our biological characteristics as well as our psychological and social ones. Even conditions such as cystic fibrosis or sickle cell anemia, which follow predictable patterns of inheritance, can exhibit a wide range of symptoms that differ in their severity in different people or, indeed, in the same person at different times.

When it comes to the more common and prevalent health conditions or diseases, such as the various cancers or the vascular conditions that can lead to heart attacks or strokes, genes do not predict the fact or time of their occurrence or their severity with any degree of accuracy. For these sorts of conditions, a person's life circumstances, beginning at birth (or indeed during gestation) are better predictors than their genes are. This is not to say that "genes" aren't involved. DNA is involved in everything that goes on in our bodies by virtue of the fact that DNA specifies the composition of proteins and different proteins are continuously being synthesized and participate in all our biological functions. In fact, that's why DNA is important. But the relationships between our DNA and our proteins are neither simple nor one-on-one and they change over time. That is what makes genetic predictions problematic and unreliable.

So let us look at a few specific examples and let us stick with the rarer conditions, the ones that follow predictable patterns of inheritance, to illustrate how little "knowing our genes" can help us even with those. The best understood inherited disease and the one about which biologists and biochemists have known for the longest time is sickle cell anemia (SCA). Its molecular nature was described in the late 1940s at which time its pattern of inheritance within families was already known. It was also known to be more prevalent among people of sub-Saharan descent. (In fact, SCA is generally believed to have become established there because it protects people who inherit the corresponding DNA sequence—the "sickle cell gene"—from malaria.)

For about fifty years, scientists have known the structure of the protein responsible for SCA. For more than a decade, they have also known the DNA sequence of the gene that specifies its

composition. It is hard to imagine knowing more about a gene and the protein to which it is linked. The point I want to make here is that none of this information has helped physicians to relieve the symptoms of SCA, much less to cure it. Measures have been devised that make it easier for some people to live with SCA, but knowing the gene is not involved. I stress this fact because of the constantly reiterated promise that understanding the relationship of genes to even complex conditions will help not only to diagnose them, but to prevent and cure them.

Just two more examples of the difficulties involved in translating knowledge about genes into predictions of disease. These, too, are relatively rare conditions whose patterns of inheritance are more or less predictable within families. One is called beta-thalassemia and, like SCA, it results from changes in the function of the blood pigment hemoglobin. Beta-thalassemia is a more debilitating condition than SCA and, in contrast to SCA, which results from only one alteration (or "mutation") in the hemoglobin gene, many different mutations in this gene can provoke beta-thalassemia. Furthermore, different degrees of disability are associated with different mutations and the degree of disability is different for different people as also for the same person at different times. Stranger yet, a mutation that elicits severe symptoms in one person may elicit no symptoms at all in others. No one yet understands why this is so except to assume that the relevant genes and proteins engage in multiple interactions with other things going on within the organism as well as outside it.

A final example to illustrate the unpredictability of the degree and manner in which gene mutations are expressed as disease. This one concerns a group of diseases of the retina of the eye, called retinitis pigmentosa, or RP, that result in a progressive loss of night vision and often lead to blindness. RP, too, is a "simple" inherited condition in the sense that it runs in families and manifests relatively predictable patterns of inheritance. What I want to stress, from the point of view of the complexity of relating genes to disease and the resulting unreliability of predictions even in the case of these relatively "simple" patterns of inheritance, is that different mutations appear to be able to elicit similar manifestations of RP. At the same time, the identical mutation, occurring in two siblings, has been known to produce blindness in one while the other, even though older, continues to be able to drive her car at night. More surprising yet is the fact that, in some forms of this inherited condition, the visual cells in one part of the retina deteriorate, so that that part of the eye becomes blind, whereas elsewhere in the same eye, the cells appear to be unaffected.

The complexities that emerge upon closer examination of such relatively well understood patterns of gene function make a mockery of the pretense that we can use the worldwide

distribution of the genes of human populations to shape our understanding of our history or to guide social policy.