

How Can Understanding Genetic Racial Distinctions Be Helpful?

What is the genetic basis of race? Are Africans genetically different from Asians? Are there race-specific genes?

The more closely researchers examine the human genome the more most of them are convinced that the standard labels used to distinguish people by race have little biological meaning. Although it may seem easy to tell at a glance whether a person is Caucasian, African or Asian, when geneticists probe beneath surface characteristics and scan the genome for DNA hallmarks of race, that seemingly obvious conclusion disappears. Humans have spread out over the world in a relatively short time. Therefore there has simply not been enough time for the human species to divide itself into separate biological groups in any genetically significant way.

Please note: definitions of terms that are in boldface in the text as well as other terms can be found in the Glossary.

Ninety-nine per cent of the human genome is similar. Within this similar portion, about 75% of all the genes come in only one allelic (monoallelic) form and are identical in everybody. Using the blood typing example explained earlier (Genetics 101, Practical Implications of Genetic

Variation: The Story of Blood Types), such single allelic genes would be like having a blood type consisting of only the A allele, which would mean that everyone would have type AA blood. Because of this 99 % genetic similarity, individual variations among human beings are accounted for by the remaining 1%. Within that 1% of genomic variability, 85% exists within any local population, be they Italians, Kurds, Koreans or Inuit. Of the remaining 15%, about 7% variability can be found within any given continent while the remaining 8% in variability occurs between large groups living on different continents. That means two random Koreans may be as genetically different as a Korean and an Italian and there is not much additional variation within a continent compared to that between continents.

The way we measure human variation genetically is to look and find all the different allelic variations of a gene and then see what percentage of each variant of that gene occurs *within* populations and *between* populations. To illustrate this, let's look at blood types again. To examine genetic differences in blood types between two populations, start by examining the percentages of the allelic variations in those two populations. For example, the distribution of blood types among people from the Philippines is nearly identical to the distribution of blood types among the population of China. There is genetic variation within each population but not much variation between the two populations. Alternatively, 100% of Peruvian Indians have blood type

O, while among Blackfoot Indians, 82% of individuals have blood type A and 18% have blood type O. Within each of these two populations, there is little or no genetic variation, but there is a great deal of variation between the two populations.

Generally, most genetic variability can be found within populations but different populations can have very different degrees of variability. In fact, about 93% of all of the genetic variability that exists on this planet occurs within Sub-Saharan Africans. So, if there were a catastrophe that destroyed the rest of the world's population, 93% of the genetic variability in the world would still be present in Sub-Saharan Africans.

Medical Implications of Racial and Geographically-distinct Human Groupings

Sickle Cell Anemia Again

Are there genetic differences between human groups that are medically important? If race has any bearing on health at all, it may simply be a marker for the geographic origins of certain populations. In the Eastern Hemisphere, where it is thought humans have lived for at least 2 million years, differences that developed in skin colour were closely correlated with latitude and exposure to sunlight. The same pattern is not apparent in the Western Hemisphere, to where anthropologists suggest humans migrated only about 35,000 years ago.

This kind of knowledge can be helpful in understanding diseases such as sickle cell anemia. This disease is often found in African and Mediterranean peoples but also among immigrants or ancestors of immigrants from these regions to North America. The higher prevalence in these peoples is thought to be at least partly due to a health advantage for persons with the sickle cell anemia in battling malaria, which is endemic to those areas. Malaria parasites do not survive as well in sickled cells and therefore those infected by mosquitoes with the parasite that causes malaria may not develop malaria or may have a milder form of the disease. Sickle cell anemia is rarely seen in descendants of people from northern Europe, where malaria is rare or absent.

Genetic Ethnic Distinctions and Other Common Diseases

So the genetic link between sickle cell anemia and protection against illness caused by malarial parasites is clear-cut. But for the major diseases that cause or contribute to most deaths and disabilities, the genetic contribution is harder to pinpoint. For these diseases, such as heart disease, high blood pressure and cancer, multiple genetic mutations are thought to increase the susceptibility of some individuals, but environmental factors, such as diet and lifestyle, also play an essential role in

development of these diseases.

It's much harder to make the case that high blood pressure is a bigger burden in some ethnic groups because of their genetic makeup. For example, high blood pressure more severely affects people of African descent in Canada compared to those of European descent.³ African-Canadians are also much more likely to die of stroke than Canadians of European descent. Some have speculated that African slaves who were better able to retain salt were more likely to have survived the deprivations of diet and sanitation on the slave ships transporting them to North America. If this were true, then the same genetic makeup that helped them survive, when passed on to their offspring, may have put later generations of black Canadians at risk for developing high blood pressure (also called hypertension). But there are also a number of societal and cultural factors that might predispose African-Canadians to hypertension including the stress of living in a prejudiced society, lack of access to health care, poor diet, etc. Complicating matters is that no one really knows which combination of genes is responsible for susceptibility to hypertension. It's likely that a large number of mutated genes may contribute to high blood pressure, but that not all patients may have all those mutations.



³ Brophy, Kathleen Marion, Scarlett-Ferguson, Heather, and Webber, Karen S. *Clinical Drug Therapy for Canadian Practice*, Lippincott Williams & Wilkins, 2010, p. 779.