III. Genetic Research in Racial and Ethnic Groups

Scientists have investigated the connection between genetic disorders genetic and racial and ethnic groups. Even before the discovery of DNA in 1953, geneticists had observed that certain single-gene disorders occurred predominately in specific racial and ethnic groups. For instance, in the early 1900s sickle cell disease was observed to occur predominately in African Americans, while Tay-Sachs disease occurred predominately in Ashkenazi Jews. These early observations meshed with existing scientific views of races as inherent biological divisions in humanity. This led many researchers to conclude that genetic disorders like sickle cell and Tay-Sachs diseases were unique to the groups in which they were most common.

In the years following World War II scientists reexamined the connection between race and genes. Research in the new field of population genetics found that no single genetic alleles were unique to any particular racial or ethnic group and that on a genetic level it was impossible to distinguish between different groups. Instead of racial and ethnic groups, population geneticists advanced the concept of the population as a more accurate representation of how genes vary between human beings. A population is a group of people that share similar variations in their genes. No population has genes that are exclusive to it however, nor do populations correspond well to commonly-defined racial and ethnic groups.

Despite this change, many single-gene disorders have continued to be identified and studied within specific racial and ethnic groups. Medical researchers have also continued to use racial and ethnic categories to study the distribution of common diseases such as cancers and heart disease. Over the past decade, as genetic research on these
common diseases has increased, many geneticists have begun to investigate whether
differences in the racial and ethnic distribution of some such diseases is the result of
genetic differences between groups. The notion that there exist genetic differences
between groups and that these differences affect health is controversial. Some scientists
believe that there exist small amounts of genetic variation between different racial and
ethnic groups and that this variation may affect a group’s risk for certain diseases. Other
scientists believe that it is social conditions, such as education, poverty and diet, that best
account for a group’s risk for disease.

Geneticists look to study racial and ethnic groups for a number of different
reasons. Often a group is targeted because a particular disease or disorder occurs at high
levels in members of the group. For instance, many studies investigating the genetic
basis of diabetes are conducted in Native American and Alaska Native tribes where the
incidence of the disease is high. In such cases researchers may target the group in order
to best address a pressing health need. They may also target the group to increase the
chance that a disease gene will be identified. In an association study, the likelihood of
finding a significant correlation between a gene and a disease is increased by the risk
level of the study participants. In general, the lower the risk that a disease gene confers
the more difficult it is to detect a significant association and thus the greater the risk level
or the larger the size a study population must be.

Sometimes geneticists may target a racial or ethnic group even if its members do
not have a high risk for a disease. The accuracy and efficiency of some genetic studies
can be improved when the study population is genetically similar, or homogenous. Some
ethnic groups, particularly those that are small in size or have historically been isolated,
may be more genetically homogenous than the general population. In association studies this homogeneity can make it easier for researchers to locate disease genes. Genetically homogenous groups are also likely to carry a smaller number of mutations in any single gene. For example, while there have found thousands of different mutations in the BRCA1 and BRCA2 breast cancer genes, Ashkenazi Jews have been found to almost exclusively carry just three. This can allow researchers to more easily study characteristics of the mutations, such as the risk they confer, as well as develop simpler and less expensive genetic tests.

To further improve the ability of geneticists to locate and study disease genes, some countries and communities have developed population-wide genetic databases. In such programs DNA samples are linked to a computer database with an individual’s health information and family history. Using sophisticated computer programs researchers can then conduct association studies within the group. Recently, a study using a population genetic database in Iceland isolated a gene that contributes to diabetes risk.

Geneticists may also look to conduct research on racial and ethnic groups for non-medical purposes. By comparing specific DNA base pairs from members of one racial or ethnic group with those from members of another, scientists believe they can estimate how closely related the two groups are. Since changes in DNA accumulate slowly over time, the number of base pairs that differ between groups may reveal the amount of time the groups have been separated. Some believe that such research will enable geneticists to identify disease genes more successfully. It may also help inform scientists and historians about the historical migrations of different populations and the genealogy of
families. Some companies now offer services to provide this information to individuals.

Such services have been popular, for example, among African Americans, who have used them to determine to which ethnic groups in Africa they may be related.

Readings

Barrett (2005) presents an argument for the use of racial and ethnic variables in medical research.

Burchard et al (2003) argue that the benefits of linking racial and ethnic categories to genetics outweigh the risks.

Hamilton (2005) examines the use of DNA testing to trace ancestry, weighing its pros and cons including the information’s emotional effect on people and its dubious accuracy.

Kalb, Springen, Carmichael, and Macgregor (2006) discuss the use of DNA tests by individuals to trace their ancestry and by geneticists to tell the story of human origins and migration.

Kingsland (2005) examines the debate around race-specific medicine.

Lee, Mountain, and Koenig (2001) discuss the consequences of using racial categories in health research.

Lemonick, Gibson, and Park (2006) provide an overview of the ongoing genetic research in Iceland by deCODE Genetics, including the scientific rationale for studying isolated populations.

Miranda (2006) discusses the boom in personal DNA tests to trace ancestry, and how the knowledge gained may challenge one’s racial or ethnic identity.


NIH, National Human Genome Research Institute, Social, Cultural and Religious Issues in Genetic Research [http://www.genome.gov/10001848] contains a discussion and references relating to race and ethnicity and genetic research.


Nature Genetics, Genetics for the Human Race (Supplemental Issue, November 2004) [http://www.nature.com/cgi-taf/dynapage.taf?file=/ng/journal/v36/n11s/index.html], although a bit more technical than other works in this area, presents interesting “state of the art” discussions of human genetics.

Nogrady (2006) provides an overview of the social and scientific controversies surrounding the issue of varying racial responses to medication.
Further Readings


Damon (1969) argues for a continued biological view of race and ethnicity in medical research.

Dobzhansky (1951) and Cavalli-Sforza (1966) provide representative scientific arguments for the replacement of race with population.

Kaufman and Cooper (2001), Kittles and Weiss (2003), and Serre and Paabo (2004) argue that human genetic diversity is best represented as a continuum.

Krieger and Fee (1996) discuss the ways in which racial and ethnic categories were applied in the efforts to collect health data.

McBride (1991) writes on how early 20th century physicians linked race and disease contagion in order to facilitate segregation.

Montagu (1962) presents an early call for the use of ethnicity instead of race in science.

Oppenheimer (2001) examines current debates surrounding the use of ethnicity versus race in medical research.

Provine (1973) and Sofair and Kaldjian (2000) provide an overview of the effect of early 20th century views of race on the development of eugenics policies.

Reardon (2005) and Marks (2000) provide an overview of the emergence of population genetics and the new discipline’s claims about race and human genetic diversity.