GENES & POPULATIONS

National Institutes of Health
National Institute of General Medical Sciences

After decades of research, scientists have identified all of the genetic "letters" that spell out the "words" of our genes. This library of genes—called the human genetic code, or the human genome—is the complete set of instructions for creating you or me. But scientists' ability to read the human genetic code is only a beginning. It will be many more years before researchers figure out what all of our genes do. This information holds tremendous promise for treating, curing, and preventing diseases. By participating in genetics research, you can help scientists understand how our genes influence the diseases we develop.

Q: Does everybody have the same genes?

A: Yes. Most genes are exactly the same in all humans, but each of us has a very small number of genes that are spelled slightly differently. These tiny differences make up much less than 1 percent of each person's genetic material (DNA), yet the differences are enough to create people with different appearances and different health. The gene differences are usually inherited, so the more closely related two people are, the more similar their DNA is likely to be.

Q: What does it mean when someone says you have a genetic risk?

A: It means that you have inherited the tendency to develop a certain illness. Having a genetic risk does not mean that you will develop a particular condition, but rather that you have a higher chance of developing it than if you did not have the risk. Your genes play a big role in determining whether or not you will develop certain illnesses. If, for instance, your mother's father had a history of heart disease, you may have inherited that genetic risk from genes you got from your mother (that she got from her father), much as you may also have gotten your grandmother's straight or curly hair. But genes are not the only thing. What you eat, whether and how much you exercise, and where you live and work are all factors that can profoundly affect your overall health.

Q: What is genetics?

A: Genetics is the study of inheritance. Genes are the instruction manuals for making building blocks called proteins, and proteins combine in many ways to create all the parts of our bodies. Proteins also create unique body features called "traits." Some traits are visible, like eye color. Others are not, like your body's ability to fight certain diseases. Scientists who study genetics are called geneticists.

Q: How do scientists learn how genes play a role in our risks for fighting or developing disease?

A: Your tax dollars, through the National Institutes of Health, fund medical research in the United States. The National Institutes of Health provides some of this money to scientists at universities and medical centers to carry out research to identify the roles of genes in various diseases. In the laboratory, genetics researchers often use organisms that can be bred easily and quickly, like plants, fruit flies, or mice. Scientists breed slightly different versions of these organisms and watch the outcomes. These experiments give scientists lots of basic knowledge about genes and diseases, because humans have many of the same genes as these experimental organisms. Knowledge about genes involved in diseases helps scientists find ways to treat, cure, or prevent diseases.

Q: How do scientists find genes linked to human diseases?

A: Researchers link genes to diseases in two different ways:

Studying a disease. Scientists who are looking for relationships between genes and a disease often start with the disease. For example, since some forms of breast cancer run in families, researchers might suspect that one or more genes plays a part in causing breast cancer in those families. To test that idea, scientists would ask family members to participate in a genetics research study so that researchers could search for a telltale gene spelling or combination of gene spellings present only in people who have breast cancer and not in their family members who don't.

Studying genes. Sometimes, earlier research in laboratory organisms has yielded a lot of basic information leading scientists to suspect that certain genes and the proteins they produce

might be linked to a particular disease—heart disease, for example. For the most part, researchers have already figured out which proteins are important for the proper functioning of the heart. For example, some of these proteins affect the pumping of the heart or regulate cholesterol levels in the blood. Scientists analyze the spellings of the genes that make those proteins and then conduct studies with DNA from large numbers of people, hoping to find certain gene spellings—or combinations of spellings—that turn up over and over again. These spellings could turn out to be predictors of heart disease.

Q: Why do researchers sometimes study ethnic and racial groups?

A: Even though everyone's DNA is just a little different from everyone else's, we all have 99.9 percent of the same genes! However, since 3 billion chemical "letters" string together to make up our genes, the one-tenth of 1 percent of DNA that is different can add up to hundreds of thousands of differences—most of which have no effect at all. Groups large or small who share the same ancestry are likely to have a greater number of similar gene spellings than do more distantly related people. Studying the DNA from family members—or from populations in which people share a common ancestry—reduces the "background noise" among all those tiny genetic differences. This makes it easier for scientists to pick out the gene differences, or patterns of differences, linked with disease. Researchers study genes from people in different population groups to find the common genetic differences that are unique *to certain diseases*, not to particular ethnic or racial groups. Occasionally, however, scientists do study members of certain ethnic groups to make it easier to identify genetic differences that are common to those groups. The gene that is misspelled in Tay-Sachs

disease, for example, was identified in Ashkenazi Jews.

Q: Have researchers figured out how errors in genes can cause disease?

A: Yes, but only for some diseases. Scientists believe that certain diseases, like cystic fibrosis, are caused by an error in a single gene. But genes alone rarely cause disease. Many genetically complex disorders that affect millions of people—for example, heart disease, depression, and cancer—are caused by many different genes. Combinations of these genes, along with how we live our lives—what we eat, whether we exercise or smoke, and what we are exposed to in the environment—cause these diseases. Researchers do not yet know what most of these combinations of factors are. To discover which genes contribute to complex diseases, scientists must keep looking to find all of the gene differences that exist in all people.

Q: What's involved in this sort of genetics research study?

A: Your body is made up of individual units called cells. Your DNA (your personal genetic code) is tucked inside each of your cells. If you agree to participate in a genetics research study, scientists will collect a small sample of your cells in order to read this code. One of the simplest ways to do this is to collect a strand of hair or a sample of blood, or to gently rub the inside of your cheek with a cotton swab. Scientists will study the DNA from these samples and, through laboratory techniques, they will read the spellings of your genes.

Q: Can researchers study my genome without my knowing about it or giving my permission?

A: No. Researchers take great care with DNA samples obtained from research volunteers.

Before participating in any research study, you are required by law to read and sign an "informed consent" form. Before collecting samples of your DNA, scientists must tell you the purpose of their study and also if—and for how long—your DNA sample will be stored in a facility called a "repository." Researchers must also get your permission to permit your DNA sample to be used by other scientists for future studies.

Q: What are the benefits of genetics research to the groups and populations whose members volunteer to participate?

A: Studying genes from people who are members of a given population or group—sometimes called a "community"—can be a good thing for that community as a whole. Research results could ultimately point to treatment or prevention strategies that benefit many people in that community.

Q: What are the risks?

A: Research results could lead to the stereotyping of a community as being more likely to have a certain disease. But this may not be the case if the reason that the disease was found in a group is that the group members were simply more willing to take part in a genetics research study.

Q: How do researchers protect people who volunteer to take part in genetics research?

A: Participating in any research study is voluntary, and there are strong safeguards to protect medical research volunteers. Scientists who conduct research with people are required to take important precautions with human DNA samples—for example, removing identifying information and not sharing the samples with other researchers without receiving permission from you. Before you agree to participate in a study,

read about the study's goals and ask the researchers what benefits and risks may result. Take time to read and understand the informed consent document that you are required by law to sign before participating in any research study. Find out which members of your community were on the "Institutional Review Board" that by law is required to review and approve all federally funded human research studies.

Q: Are there other protections for the DNA samples collected by researchers?

A: One example of other protections is found at the National Institute of General Medical Sciences Human Genetic Cell Repository. The National Institute of General Medical Sciences (a part of the National Institutes of Health) provides funding and oversight to this repository. Scientists can obtain samples from the repository to use for their genetics research studies. Cells stored in the repository are not identifiable by donor. When you agree to participate, scientists should also provide you with information on how you can learn about the results of research studies performed with your DNA sample. To protect identified ethnic and racial communities, the National Institute of General Medical Sciences requires that researchers who wish to collect tissue samples from members of communities must first talk to community members and community leaders.

The National Institutes of Health is dedicated to listening to both individuals and communities about the impact of genetics research. The National Institutes of Health held the first "community consultation" on the topic in September 2000. An ongoing dialogue with communities has only begun. More discussions with various communities have been held or are being planned.

For more information, or to learn more about how you or your community can get involved, contact the National Institute of General Medical Sciences Office of Communications and Public Liaison (301-496-7301) or visit the World Wide Web sites below.

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