

The Haplotype Map Project (HapMap) and Other Research on Genetic Variations

Please take as much time as you want to read this form, ask questions, and talk about this project with family or friends.

What is this project about?

The National Institutes of Health (NIH), the main U.S. government agency that funds medical research, is collecting blood samples from families whose ancestors came from different parts of the world. About 100-250 people from each of about 10 ethnic or geographic groups will give samples for this project. The University College Hospital (UCH) is helping the NIH by asking families in your community to give samples. We will not collect any medical information, but researchers who study the samples will know which ethnic or geographic group each sample came from.

Why is the NIH doing this project?

Genes are the basic “instruction book” for people. Genes are made out of DNA. The DNA of a person is about 99.9% the same as the DNA of any other person. But no two people have exactly the same DNA except identical twins. Differences in DNA are called genetic variations. They explain some of the differences among people, like eye colors and blood groups. They also partly explain why some people get diseases like cancer, diabetes, asthma, and depression, while others do not. Diseases such as these are also affected by diet, exercise, smoking, pollution, and other factors, which makes it hard to figure out which genes affect the diseases.

Most genetic variations are found in people everywhere. But there are differences among groups in how common some genetic variations are. For example, all the blood types can be found in all groups of people in every part of the world, but there are differences from place to place in how often some blood types appear. By including families from many ethnic and geographic groups, researchers will find most of the genetic variations. This information will make it easier for researchers to find the genes that affect diseases, in other studies using other samples.

How will the samples be used?

Over the next three years, researchers will study the samples to find places in the DNA where people vary. For each sample, researchers will make a list of the genetic variations they find. Researchers will also look for the patterns of genetic variations in people’s DNA, which are called “haplotypes.”

Researchers will put all this information in a scientific database on the Internet. For each sample, this will include information on hundreds of thousands (eventually millions) of genetic variations, as well as the ethnic or geographic group, the sex, and the position in the family of the person who gave the sample. The database will not include any medical information about anyone who gave a sample. It also will not include any information that could identify who the individual people or families are.

Researchers will use the genetic variation information in the database to create a genetic map that summarizes the patterns of genetic variation, called a haplotype map or “HapMap.” The HapMap will be put on the Internet. The HapMap will not include medical information, but researchers will use it as a tool in future studies to find genes related to many diseases. The HapMap will show researchers where the haplotypes (patterns of genetic variation) are. Then, for a disease, such as diabetes, researchers will

study the haplotypes in a group of people who have the disease, and in another group of people who do not. Areas in the DNA where the two groups differ in their haplotypes will be clues that those areas might contain genes that affect the disease. Researchers can then look for those genes and study how they work. This will help them figure out better ways to prevent, diagnose, and treat the disease. They can also learn how to make drugs that work better in more people. Some researchers will also use the HapMap to look for genes that affect traits such as baldness, behaviors like addiction, and long life.

Researchers will compare the genetic variation and haplotype information for people in the same group and in different groups. In the future, researchers will also use the samples to look for differences in the amount and form of the products that genes make, called RNA and proteins, and will put all this information for each sample in the database. The samples, the database, and the HapMap will also be used to study other questions, such as the biology of DNA, how new variations arise, the genetic history of human groups, and how people from different parts of the world are related to each other.

Because the database will be public, people who do identity testing, such as for paternity testing or law enforcement, may also use the samples, the database, and the HapMap, to do general research. *However, it will be very hard for anyone to learn anything about you personally from any of this research because none of the samples, the database, or the HapMap will include your name or any other information that could identify you or your family.*

What will happen if I decide to give a sample?

We will ask where your grandparents came from and what language they spoke. (Tell us if you are unsure, because our samples should be from people whose grandparents were mostly Yoruba.) If you think someone in your family might not be biologically related, we would like you to tell us but you don't have to. The place we store the samples will test them and use samples only from families where everybody is related. If they figure out that someone in your family is not biologically related, they won't tell you, your family, or anyone else, since they will not know which samples came from your family.

We will also ask if your family is biologically related to any other family who gave samples for this project, since we can include only unrelated families. We may also ask how old you are to make sure that you are age 18 or older.

We will not be able to use samples from persons with HIV for the HapMap. Therefore, we must make sure that none of the samples donated for the project contain HIV. First, trained individuals from your community will provide HIV counseling for persons that would like to participate in the HapMap, then they will draw a blood sample for HIV testing. When the test results become available, each person will receive his or her results from a trained counselor during another counseling session. The HapMap investigators and staff will not be directly involved in the HIV counseling and testing. Persons with a positive HIV test will not be able to participate in the HapMap, but will be referred to local HIV clinics for treatment and follow-up care. Persons with a negative HIV test will be eligible for the HapMap project.

We will use all this information to decide whether you can give a sample, but we will not keep any of this information or put it in the database. If you meet the requirements for the HapMap, and agree to participate, we will draw about 4 tablespoons of blood from your arm for the project. We will send your samples to the Human Genetic Cell Repository at the Coriell Institute for Medical Research in New Jersey U.S.A. (the "Repository"), which the NIH oversees. They will turn the samples into cell lines, which will make an unlimited amount of DNA and will last a long time.

The Repository will send the cell lines to researchers around the world to create the HapMap and to use in many future genetic studies as described in this form. The researchers will have to follow all U.S. and international laws and guidelines that apply to research. All studies using the cell lines will have to be approved by the Institutional Review Board (IRB) of the Repository. An IRB is a committee similar to the one that approved this project to make sure that your rights were protected. Also, a Community Advisory Group will be set up for each community that takes part in this project. This group will include people from your community and will make sure that future studies using your community's samples are similar to ones described in this form. This group will also suggest ways to do those studies to limit any possible harm to your community.

Will there be any costs or payments?

It will not cost you anything to be a part of this project. We will take care of expenses related to the study, including the costs for HIV counseling and testing. We will compensate you for your time, travel, and inconvenience if you come in to give us a sample.

The Repository does not let anyone sell material from samples or cell lines. However, information from genetics research sometimes helps companies make products to diagnose or treat diseases. If information from your family's cell lines leads to making a product, it would probably contribute only in a very small way. Also, because the cell lines will not have names on them, neither the researchers nor anyone at the Repository would know if your samples were even used. So you will not get any additional payment if you take part in this project.

How will you protect my privacy?

We will protect your privacy in several ways. While UCH will keep your signed consent form, nobody else will see it. We will not keep your name with your sample or give your sample a code number that could identify you. So nobody at the Repository or who studies your sample will know that it came from you.

Also, we will collect more samples than we will use. This way nobody, not even you or us, will know if your family's samples were used or if any information in the database came from your family. (Samples that are not used will be disposed of in standard ways.)

What are the benefits of giving a sample?

You probably will not directly benefit from giving a sample, because of the long time this research will take to produce useful results. But researchers will study these samples for many years to learn about health and disease. This research will eventually benefit the health of people around the world.

What are the risks of giving a sample?

Drawing blood has very minor risks. These include brief pain, slight bruising, dizziness or fainting, and (very rarely) infection where the needle goes in.

If your family's samples are used, lots of genetic information from your samples will be put in the database, and lots of people will be able to look at it for any purpose. However, there are only a couple of ways anybody could trace the information back to you. One is if they thought your information might be in the database, got another sample from you, did many tests on that sample, and then compared the genetic information from those tests with the information in the database. The other is if somebody

compared the information in the database with genetic information known to be from you that was in another database and figured out who you were. The risk of either of these things happening is very small, but it may grow in the future.

We cannot always predict the results of research, so new risks to you may come up in the future that we can't predict now. Your sample will not be used to make a clone of you.

Are there any risks to my community or group?

Information on the ethnic or geographic groups the samples came from will be included with the samples, in the database, and in the HapMap. In future studies, researchers may find that certain genetic variations appear more often in people from your group than in people from other groups, and that these variations are more common in people with a certain disease. This may make some people look down on your group unfairly.

Some people may use the information from the HapMap or from future studies using the HapMap to exaggerate differences between groups for prejudiced or other bad reasons. Others may use the information to downplay differences between groups, to say that all people's genes are about the same, so we don't need to respect the special concerns of different groups. Biology does not provide a reason for prejudice, but discrimination does exist.

We will work to make sure that the ethnic or geographic identity of your community is described as carefully as possible--in the sample collection, in the database, in the HapMap, and in any articles researchers write about the HapMap.

Can I change my mind after I give a sample?

Giving a sample is completely up to you. You will not lose any benefits if you choose not to give a sample. However, since nobody will know which sample came from you, after you give a sample you cannot take it back or take any information out of the database.

How will I find out what happens with this project?

Because your sample will not have your name on it, we will not be able to give you individual results from this research. However, we will update your community through your Community Advisory Group on how researchers are using the HapMap and your community's samples and what they are learning about health and disease.