

Sample Consent Document 7: 1000 Genomes

Project: Developing a Research Resource for Studies of Human Variation

This consent document was used to recruit participants for a whole genome sequencing study. It includes language about the sharing of sequence data through open access.

Important note: This consent document was developed for the 1000 Genomes Project. It is not provided as guidance or as a template promoted by NHGRI, but as a reference to inform investigators and IRBs considering these issues. It is important to tailor consent documents for each individual study.

1000 Genomes Project: Developing a Research Resource for Studies of Human Genetic Variation

CONSENT TO PARTICIPATE

1. *Introduction*

We invite you to be part of the **1000 Genomes Project**, which will develop a research resource that researchers around the world will use. This resource will be a catalog of human genetic variation, and will include both: (1) blood samples and material taken from the blood samples, which will be kept in a repository and distributed to researchers for use in future projects; and (2) data from the study of the samples, which will be kept on scientific databases available over the Internet. The resource will be used in many future studies related to health and disease.

Researchers in several countries are working together to develop this resource, including [institution]. Several agencies are sponsoring the project, including [list]. You can learn more information about the organization of this project at <http://www.1000genomes.org>.

This project will involve at least 1,000 men and women whose ancestors are from various parts of the world. [Institution] is recruiting some of these participants. In order to take part, you must:

- be at least 18 years old [or applicable age of majority];
- be willing to give a sample of blood so that researchers can read out all of your genetic information from it (a process called “sequencing”);
- be willing to have a “cell line” made from the blood sample that will make it possible for researchers to get an unlimited amount of genetic material from the sample for a long time—maybe forever;
- be willing to have all of your genetic information (without your name or other traditional identifying information, such as address, birth date, or [U.S. only] Social Security number) put in scientific databases available on the Internet;
- be willing to have many researchers around the world study the genetic material and data from the sample for a long time, and to have the information they learn put in scientific databases on the Internet.

We will not collect your name or any medical information. Researchers who study the material and data from the samples will be told only the sex of each donor and which ethnic or geographic group the donor came from.

If you think you might want to be part of this project, please read the rest of this form and take as much time as you need to ask questions.

The decision about whether to participate or not is completely up to you. [The following sentence is optional but may be required by some IRBs in the U.S.] If you decide not to sign this form, it will not affect any benefits to which you are entitled.

2. *What is the purpose of this research?*

Genes are the basic “instruction book” for the cells that make up our bodies, and are made out of DNA. The DNA of a person is more than 99% the same as the DNA of any other unrelated 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 physical differences among people, and partly explain why some people get diseases like cancer, diabetes, asthma, and depression, while others do not. Such diseases may also be affected by factors like diet, exercise, smoking, and pollution in the environment, which makes it hard to figure out which genes affect the diseases.

Most genetic variations are found in people across the world. But there are differences among groups in how common some genetic variations are. **The purpose of the 1000 Genomes Project is to find most of the genetic variations that exist in people. We will do this by studying the DNA in blood samples collected from many people whose ancestors were from various parts of the world, and then putting all of this information in scientific databases on the Internet.** These scientific databases will be kept for a long time, and many future researchers around the world will use them to help find genes and genetic variants related to health and disease.

The scientific databases we develop for this project will not include any medical information, but they will still be useful to help future researchers learn about health and disease. In the future, for a disease (such as diabetes), researchers will study different sets of samples—some from people who have the disease and some from people who do not, and look for areas in the DNA where the patterns of variation differ between the two groups. This will give them a clue that those areas might contain genes that affect the disease. They will then use the scientific databases we develop for this project to look at the genetic variants in those regions, to help figure out which genes might affect the disease. They can then study how the genes work and eventually find better ways to prevent, diagnose, and treat the disease. Researchers will also use the scientific databases to learn more about how different people respond to different drugs, and about how traits (like long life), or behaviors (like addiction), differ between people. Other future studies that use scientific databases and the samples themselves will help researchers understand even more about human genetic variation and other important biological questions.

This is a research project, not medical care. You should see your health care provider for any scheduled visits or if you have a health problem or medical question.

3. *What will you ask me to do if I want to participate?*

We will first ask you a few questions to determine whether you are eligible to give a sample.

- We will ask you how old you are, since only people who are [age of majority] may contribute a blood sample.
- We will ask you where your grandparents were born, and [where appropriate] what language they spoke. Tell us if you are unsure, because the samples should be from people whose grandparents mostly came from [geographic location or ethnic group].
- We will ask you whether any of your relatives already gave samples for this project. We need to collect samples from people who are not closely related to each other, so we do not want to include more than one member from a family.
- [may need to include additional topics here, as applicable]

If we find that you are eligible to give a sample, we will then draw about [amount] of blood from your arm.

4. *What will happen with the sample I give you?*

We will send the sample to the [Repository] to be processed and stored. [Repository] is located in [city, state/country] and is overseen by [agency]. **The only information we will include with the sample is the name of the ethnic or geographic group you come from (or that your ancestors came from), and your sex.** [May need to modify this in some cases.]

[Repository] will:

- make a “cell line” from the sample so that researchers can get an unlimited amount of DNA from it for a long time—perhaps indefinitely; and
- send the DNA from the cell line to project researchers in laboratories around the world.

Over the next 1-3 years, these project researchers will:

- “sequence” the sample, by reading out all of the genetic information in it;
- put all the data in open access (public) scientific databases on the Internet (at [cite URLs for website(s)] through which the data can be accessed);
- study the genetic variation data from all the samples; and
- compare individual samples and samples from different ethnic or geographic groups.

[Repository] will also make the cell lines and DNA from the samples available to other researchers around the world, so that they can be used in many future studies. These future researchers may include researchers in universities, hospitals, non-profit groups, companies, and government laboratories. Such researchers, just like the researchers in this project, will have to follow all the laws and guidelines that apply to biomedical research.

Future researchers may use the samples to study many other questions, such as how genes and genetic variants affect the way genes work and the products that genes make (these are called “gene expression” or “proteomic” studies). Future researchers may also use both the samples and the scientific databases to study such questions as:

- the biology of DNA;
- how new variations arise;
- how the process of evolution works;
- the composition and size of human groups; and
- how people from different parts of the world are related to each other.

People who do identity testing, such as for paternity testing or law enforcement, may use the samples and the scientific databases to do general research about patterns of human genetic variation. Some of the results of that research, and of other future research using the samples or the scientific databases, may also be put in open access scientific databases on the Internet. However, it will be hard for anyone to find out anything about you personally from any of this research because the samples and the scientific databases will not include anybody’s name or other traditional identifying information (such as address, birth date, or [U.S. only] Social Security number).

[Note: this next paragraph may need to be modified, depending on the procedures that are used at the repository where the samples will be stored, and the last two sentences are optional, to be used only where a Community Advisory Group will be used.] Any future researcher who wants to study your sample will be required to apply to [Repository] with a written “Statement of Research Intent.” [Repository] will review each Statement of Research Intent to make sure that the purpose for which the sample will be used

is consistent with this consent form. In addition, a Community Advisory Group will be set up for your community. This group will include people from your community and will receive regular reports from [Repository] that describe how the samples are being used.

5. *What are the costs and payments?*

It will not cost you anything to participate in this project. We will compensate you [amount] for your time, travel, and inconvenience if you come in to give us a sample.

[Repository] does not allow anyone who receives DNA or cell lines from [Repository] to sell them. [Note that this is true for some repositories, such as the Coriell Institute, but may not be true for all; this sentence may thus need to be deleted.] However, some of the research done with your samples or the information in the scientific databases may eventually lead to the development of new predictive or diagnostic tests, medicines, or other commercial products. If this happens, you will not receive any of the profits from those products, or any discounts on or special access to the products.

6. *Are there any benefits to participating in the project?*

You probably will not benefit personally from giving a sample for this project because this kind of research usually takes a long time to produce medically useful results. However, your participation will help researchers around the world understand more about human genetic variation and how it relates to health and disease.

7. *What are the risks of participating?*

You may have some brief pain and bruising when we draw your blood. There is also a small chance that you may get an infection, have excess bleeding, become dizzy, or faint from the blood draw.

Although we will not collect any names or medical information, and we will take many measures to protect your privacy (see #8. *How will you protect my privacy?*), we will generate lots of genetic information about each person whose sample is studied. This information will be put in open access scientific databases, available on the Internet to anyone who wants to look at it. Although only experts will know how to interpret this information, there is a small chance that somebody could figure out how to connect you with the information from the study of the sample you give; the information could then be used to discriminate against you or your family members. Currently, we believe this could happen only if somebody knew that you had given a sample to be studied for this project and:

- got another sample from you, found an expert to test that sample, and then compared the genetic information from that test with the genetic information in the scientific databases;
- found an expert to compare the genetic information about you in the scientific databases with information known to have come from you (or from a family member) included in some other database developed by someone else for some other purpose; or
- found an expert to look in the scientific databases for a particular genetic variation known (or someday found) to be associated with a disease or trait that you have or carry, that others know about or can see, and that is very rare.

Any of these things would require that the person trying to link the information to you knew that you participated in the project. For this reason, to minimize these risks, you may wish to limit the number of people you tell about your participation.

As technology advances, there may be new ways of linking information back to you that we cannot foresee now. Also, we cannot always foresee the results of research, so new risks may come up in the future that we cannot predict now. We believe that the benefits of learning more about human genetic variation and how it relates to health and disease outweigh the current and potential future risks, but this is something that you must judge for yourself.

If you believe you have been injured because of this research, please contact [contact person]. Decisions about payment for medical treatment for injuries relating to your participation in research will be made by [institution].

8. *How will you protect my privacy?*

We will protect your privacy in several ways:

- We will store your signed consent form in a locked file; only members of the study team at [institution] will have access to this file.
- We will not collect your name or any other identifying information (such as address, birth date, or [U.S. only] Social Security number) or give your sample a code number that could identify you.
- We will collect more samples than we will use, so that nobody—not even you or us—will know for sure whether your sample was used or if any of the information in the scientific databases came from your sample. Samples that are not used will be destroyed.

Because of these measures, **it will be very hard for anyone who looks at any of the scientific databases to know which information came from you, or even that any information in the scientific databases came from you.**

9. *Are there any risks to my community or group?*

The names of the ethnic or geographic groups the samples came from will be included with the samples and in the scientific databases. 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 scientific databases, or from future studies using the scientific databases, 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 scientific databases, and in articles that project researchers write based on this research, but we cannot completely control how this information is described in publications that others write.

10. *Can I change my mind after I decide to participate?*

Because we will not collect any names with the samples, we will not know the identity of the person who gave any particular sample. For this reason, once you give a sample, it will be impractical for us to

withdraw it from the project. Also, once the sample has been studied, you cannot take the information about the sample out of the scientific databases.

[The following sentence is optional, to be used only where a Community Advisory Group will be used.] The Community Advisory Group set up for your community may request that the entire community's samples be withdrawn from [Repository], if they find that the samples are being used for future studies that are inconsistent with those described in this consent form and such studies are not acceptable to the community.

11. How can I find out about the results of the research?

It will probably take a long time for the data from this project to be used to produce health-related information that we will know how to interpret accurately. For this reason, and because we will not know who the individual sample donors are, we will not be able to give you individual results from this research. However, you can check the web sites at [cite URLs for relevant websites] to see what the uses are, and eventually what sorts of results have been found. [The following sentence is optional, to be used only where a Community Advisory Group will be used.] Also, we will update you through your Community Advisory Group on how researchers are using the scientific databases and your community's samples and what they are learning about health and disease.

The 1000 Genomes Project: Developing a Research Resource for Studies of Human Genetic Variation

CONSENT TO PARTICIPATE

Whom can I talk to if I have questions or problems?

If you have questions about this sample collection, contact:

(PI) _____ (phone) _____.

If you have questions about your rights as part of this research project, contact:

(IRB) _____ (phone) _____.

[Optional; to be used only where a Community Advisory Group will be used]: If you have questions for your Community Advisory Group about your community's stored samples, contact:

(Repository) _____ (phone) _____.

Consent and Signature

Please read the information below, think about your choice, and sign if you agree.

I agree:

- to give a blood sample;
- to have a cell line made from the sample that will make it possible for researchers to get an unlimited amount of DNA from it for a long time;
- to have the cell line and DNA used in both the 1000 Genomes Project and in other studies as described in the consent form;
- to have the entire genetic code from the sample deposited in open access (public) scientific databases on the Internet;
- that the sample or the data from my sample may be studied by companies, and that if any commercially valuable products result from these studies, I will not receive any profits; and
- that once the sample has been studied, I cannot take the information about the sample out of the scientific databases.

I have read or listened to the information, I have asked any questions I had, and all my questions were answered. I know that giving a sample is my choice.

Your Signature _____ **Date** _____

Copy given to participant: _____ **Yes**