

The *Genome Russia* Project: Research of Human Genetic Heterogeneity and Genome Variation in Russia

Informed Consent to Participate in the Project

1. Introduction

We invite you to participate in the *Genome Russia* project (<http://genomerussia.spbu.ru/>) which will develop a research resource that researchers around the Russian Federation and the world will use. The overall objective of this project is to describe variations in the human genome in the genetic heterogeneity of the Russian population, to determine characteristics that influence spread of diseases, as well as to create a database of medically significant genomic variants specific to the population of Russia, which will be a basis for developing medically significant genome variants in future. The specific objectives of the project are: (1) to collect blood samples and DNA extracted from the blood samples, which will be kept in a repository and distributed to researchers for use in future projects; and (2) to create scientific databases from the study of the samples available on the Internet. The resource will be used in many future studies related to health and disease.

Researchers from different scientific, medical and educational state institutions in Russia including St. Petersburg State University are working together in order to undertake collective research within the framework of this project. Saint Petersburg State University is the principal sponsor of the project, and The Theodosius Dobzhansky Center for Genome Bioinformatics of St. Petersburg State University (<http://dobzhanskycenter.spbu.ru/>) is the coordinator of the research consortium established to carry out the present project. The supervisor of the project is the director of The Theodosius Dobzhansky Center for Genome Bioinformatics of St. Petersburg State University professor Stephan J. O'Brien.

The present project will include obtaining DNA from at least 2500 volunteers from different parts of Russia, whose ancestors were indigenous to the region for several generations.

In order to participate, you are obliged to:

- be at least 18 years of age;
- agree to give a blood sample of 7 ml to be kept and used in the scientific purposes;

- agree to decode the genome DNA sequence and keep your genetic information (without mentioning the name or other personal data) in scientific databases available on the Internet for scientific research.

The collected personal information (names, contact information, addresses) will be kept separately and will not be transferred to anybody. After the research is finished, the files with all the personal information will be eliminated. The researchers analysing the DNA from the blood samples will only be aware of the sex of each donor, the year of birth and the ethnic or geographic group the donor belongs to.

If you think you might be willing to participate in the 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. If you decide not to sign this form, it will not affect medical care you will receive in future or any benefits to which you are entitled.

2. The purpose of the research

Genes are the basic “instruction book” for the cells that make up our bodies, and are made out of the DNA. The DNA of a person is more than 99% the same as the DNA of any other unrelated person; however, 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 different factors such as diet, exercise, smoking, and environmental pollution.

Genetic variations are found in people across the world. **The purpose of the *Genome Russia* is to find most of the genetic variations that people living in the Russian Federation have.**

The data will be used for different research. The main purposes are: 1) To identify the pattern and distribution of known diseases causing gene variants across the Russian population. 2) To develop a Russian population based Haplotype map (HapMap) required to identify disease gene markers specific for high incidence Russian diseases. 3) To discover and catalogue new gene variants specific for certain Russian ethnic groups. 4) To interpret laws of the variability of human DNA to decipher historical migratory routes and settlements of the ethnic groups.

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

3. What should I do if I want to participate?

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

- You will be asked how old you are since only people who are above 18 y.o. may contribute a blood sample.
- You will be asked about the places of birth of yours, your parents and grandparents. Tell us if you are unsure about this information because the samples should be from people whose grandparents are from a single ethnic group and have been living in the same place for several generations.
- You will be asked whether any of your relatives have already given blood samples for this project.

If we find out that you are eligible to give a sample, a qualified medical worker will then draw about 7 ml of blood from a vein of your arm at the medical institution or at your home in full compliance with standards of the Ministry of Health of the capture of CBC.

In case the analysis of your DNA will show that you have highly scientifically valuable genetic variants, the research group may (if you agree) once again refer to you in order to tell this information and get additional one.

4. What will happen to the sample I give you?

1. The blood sample will be sent to the Theodosius Dobzhansky Center for Genome Bioinformatics, St. Petersburg State University, to be processed and stored.
2. The information about each blood sample will be depersonalized and each sample will be given a unique code. Personal information collected during the project will be kept in a special protected storage and will never be connected in open databases with the information about your blood sample and DNA.
3. Your blood sample and DNA will be stored in St. Petersburg State University in a freezer for future applications related to this project.

4. Over the next 1-3 years the project researchers will do one or more of the procedures listed below:

- "sequence" (decode all the nucleotide sequences) the DNA extracted from blood by reading all of the genetic information encoded in DNA;
- determine the genotype variability in your DNA;
- put the sequence data and/or genotyping of DNA in the open access through scientific databases on the Internet without any personal information;
- compare the individual samples with the samples from different ethnic or geographic groups;
- study the genetic variations of DNA samples connected with human diseases.

5. Are there any benefits of participating in the project?

The *Genome Russia* project is a nonprofit academic research and does not imply receiving any direct benefits. However, your participation in the project will significantly help to create an ethnogenetic map of Russia and produce principles of personalized medicine. What is more, in case your DNA sample is decoded, you might be able to receive a document with the list of the genetic variants in your genome, on the basis of which the medical interpretation of your genome can be made in specialized medical institutions.

6. What are the risks and disadvantages of participating in the project?

The participation in the project does not imply any risks. When taking a blood test you may feel discomfort or dizziness. You may have bruising or swelling of the arm at the injection site.

7. How will you protect privacy of my genetic data?

In the course of the study, the records relating to you will be kept confidential. A consent form signed by you and a questionnaire will be stored in protected folders; and only members of the research group, who collect the blood and are members of the *Genome Russia* project, will have access to these folders. The staff, who will carry out your DNA sequencing, processing and

producing a database, will not have access to your personal information. After the project and processing is finished, all the personal information will be eliminated.

8. Are there any risks to my ethnic group or community?

The names of the ethnic or geographic groups the samples came from will be included 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.

Some people may use the information from the scientific databases or 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 do not need to respect the individual biological features of different groups.

In the course of the project, 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, scientific databases, and academic articles that researchers of the present project will write based on this research; however, we cannot completely regulate the way the received information is described in the publications written by other scientists.

9. Can I change my mind and cancel my participation in the project?

Once the sample has been studied, you cannot take the information about the sample out of the scientific databases. However, if your sample has not yet been processed, studied and placed into a computer database, in case you change your mind about continuing participation in the project, we may remove your sample from further research by your request.

10. 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 and knowledge how to interpret this information accurately. You can check the website of Dobzhansky Center for Genome Bioinformatics (<http://dobzhanskycenter.bio.spbu.ru/ru/>), which will provide information on the progress of the project.

11. Can I have access to my own DNA sequence?

If your DNA sample is sequenced, you may request access to the data of your DNA, in case you have selected this variant when signing the present informed consent.

Contact information

If you have questions about the study, you may contact:

Brukhin Vladimir Borisovich, Leading Researcher, PhD
Theodosius Dobzhansky Center for Genome Bioinformatics
VO Sredny Prospect 41, 199004, St. Petersburg
tel. +7 (812) 363-61-03, email: vbrukhin@gmail.com

If you have questions about your rights as participant of the project, you may contact:

Ethics Committee of St. Petersburg State University
Email: irb@spbu.ru

I have attentively read the information about the *Genome Russia* project and agree:

1. To give a blood sample;
2. That my blood sample and the DNA extracted from it can be used only for the scientific purposes of the abovementioned areas. It cannot be sold or used for other purposes.
3. That my name will not be mentioned on my blood samples and the DNA extracted from it. The samples will be marked only by the code number of the research participant that will not contain any personally identifiable information.
4. That the DNA decoding results produced using my blood samples will be anonymously posted in open scientific databases on the Internet.
5. That the results will not be placed in my medical record. I cannot be personally identified in any publication related to the research.
6. That once the sample has been studied, I cannot take the information about it out of the scientific databases.
7. To report all information included in the questionnaire.

I'd like to obtain an expert opinion on my DNA sequence if my sample is selected for sequencing my genome.

Yes _____ No _____

I am fully acquainted with the text of the informed consent. I understood all the information. I voluntarily agree on storage and further use of my blood samples

Participant's name and surname _____

Passport _____

Participant's signature _____

Name and surname of the research group representative _____

Signature of the research group representative _____

Date _____

A copy of the signed consent is given to the participant of the project