COLUMBIA | CPMG

CENTER FOR PRECISION MEDICINE AND GENOMICS

Consent Statement and HIPAA Authorization

Study Title: Genetic Studies of Constitutional Disorders
Study number: AAAS7948

Anticipated number of Subjects: 13,500

Study duration: 1 hour

Principal Investigator: Ali Gharavi, MD
Contact number: 212-851-4927
Email: cpmg_info@cumc.columbia.edu

Funding Provided by: the National Institutes of Health (NIH) and the Center for Precision Medicine and Genomics (CPMG)

This form is written to address a research subject. If you are signing this form as a parent/legal guardian of a minor, please read "you" as "your child".

You are being asked to take part in a research study. This form explains why we are doing this study and what you will be asked to do if you choose to be in this study. It is important for you to understand why the research is being done and what it will involve. Please take time to read the following information carefully and discuss it with friends and relatives if you wish. Ask us if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part in this research study.

The purpose of this research is to learn more about the genetic material (DNA) of individuals with constitutional disorders. Constitutional disorders are medical conditions that require a visit to an internal medicine doctor or pediatrician. They include health problems related to the heart, lungs, kidney, liver, or any other organ or physiological system.

1. Why am I being asked to participate?

We invite you to participate in this study because you or a family member has a constitutional disorder. This study is interested in learning more about the genetic factors that put people at risk for having these disorders. Taking part in this study is voluntary and will not change your medical care.

2. What is genetic testing?

Our genetic information, or DNA, is made up of a code that provides information for carrying out all of our body's functions. Specific parts of DNA are called genes. Genes provide instructions for carrying out specific functions of the body. This study will perform diverse tests looking at the entire set of a person's DNA. These tests include whole genome sequencing and whole exome sequencing, as well as other new genetic tests that may be developed in the future. We are performing these tests to learn more about changes in DNA that cause or increase the risk of developing certain medical conditions such as heart, lung, kidney, or liver disease.

3. What does this research study involve?

- Review of your medical records and collecting your health history
- Collecting information about your family health history
- Obtaining and storing a DNA sample or genetic data. This may be from blood (up to 30 mL or ~3 tablespoons of blood for an adult, 20 mL or ~3 teaspoons of blood for a child), buccal (cheek) swab, or a saliva sample.
- Optional Samples: With your permission, we may contact you in the future to ask you to give another blood, cheek swab, saliva sample, or a tiny portion of the tissue leftover from your biopsy that occurred as part of your routine care for this study.
- Optional Future Contact: With your permission, we may contact you in the future to inform you about other studies. You have the opportunity at the end of this form to choose whether you agree to this future contact.
- Optional Return of Genetic Results: You have the opportunity at the end of this consent form to choose if you would like to receive genetic results. We also want to understand what it is like for you to receive genetic results. If you choose to receive results, we will only return genetic finding that explains your (or your family member's) condition or that increases your risk of having other medical conditions, even if it is unrelated to the purpose of the study. If you are found to have a genetic finding, we will ask you to give another sample to have the results confirmed in a CLIA *(Clinical Laboratory Improvement Amendments)* certified lab, and to complete 3 surveys about receiving the results. You can complete these surveys online and each will take ~30 minutes. The first survey will be completed prior to receiving results, the second will be completed one month post-results, and the third will be completed 6 months post-results. The study will cover the cost of CLIA testing.

4. Will I learn the results of this study?

If you choose to receive genetic results, we will contact you if we identify a genetic change that is important to your health. This may be a change that:

- Causes or may cause a medical condition that you already have (diagnostic).
- Increases the risk of having a medical condition for which you may be at risk and that, if not treated, is likely to cause early death or disability, and should change how your doctors care for you (secondary findings).

You can choose to receive or not receive either of these types of genetic changes. It is possible to identify more than one genetic change. You will not be contacted if you are not found to have one of these genetic changes.

How will I receive these results?

If you choose to receive results and are found to have a genetic change (as described above), we will contact you to schedule a visit with a genetic counselor. Before we can return results to you, they must be confirmed in a CLIA-certified lab as described earlier. At this visit, you may provide consent for CLIA testing as well as give another blood sample, cheek swab, or saliva sample. If we are not able to reach you, we will not return any genetic results to you. Therefore, please inform us if there is any change in your contact information. Additional testing or discussions with a health care provider may be important to pursue if you have a genetic change.

5. Will I benefit from participating?

In most cases, this study will not improve your health or directly benefit you. In some cases, we may discover information that may be of benefit to you and your family and we will contact you if you choose to receive results.

6. What are the risks involved?

- Blood draw: brief pain, bruising, or light-headedness. There is a small risk of infection when blood is drawn.
- Psychological risk: If you want to receive results, you should be aware that genetic research may identify genetic changes that may require additional testing to evaluate. Genetic research may identify serious, untreatable genetic conditions. Such a finding can result in unexpected psychological trauma, both for you and your family (if you decide to share results with them). The detection of such a condition could also affect the healthcare needs of your siblings, children, or other close relatives.
- Loss of privacy: Loss of privacy means having your personal information shared with someone who is not on the study team and was not supposed to see or know about your information. We plan to protect your privacy and the risk of loss of privacy is low but not zero.

7. Are there any costs associated with participating? Will I be paid to take part in this study?

There are no costs to you or your insurance to participate. You will not receive payment or other reward for taking part in this study. However, individuals who agree to give additional samples in the future and whose repeat sample visit is not at the same time as a regularly scheduled clinical follow-up appointment (those who come in specifically for this purpose) will receive a $25 gift card.

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8. What will happen to my data and biospecimens (samples)?

Your samples and / or data will be stored indefinitely at CUIMC, with the researchers on this study and in a central storage facility called a repository. Your samples and/or data will be identified by a unique code number that is linked to your name. The key to the code will be stored securely on the researchers' or repository's password-protected data server.

Your samples and/or data may be used by other CUIMC researchers or researchers at other institutions, including commercial companies, for research on constitutional disorders or other medical conditions. If they are shared with researchers who are not on this study, they will only be shared in de-identified form. This means that your name and other identifying information have been removed from your samples and/or data or that your samples and/or data are coded and the researchers who will use them will not have the key that links your name to the code number. It is possible that your genetic information, when combined with information from other public sources, could be used to identify you. We believe that this is unlikely to happen.

9. How will my private information be protected?

Access to your health information is required to be part of this study. By participating in this study, you are giving us permission to use the protected health information that can identify you. We will do everything we can to keep your information private. However, we cannot guarantee total privacy. Information that could directly identify you will never be shared outside of CUIMC and New York Presbyterian Hospital (NYPH), unless we are required by law, or you have given permission to share the information. The following people / groups will be able to look at, copy, use, and share your research data:

- The investigator, CUIMC and NYPH study staff, and other professionals who may be evaluating the study
- Authorities from CUIMC and NYPH, including the Institutional Review Board
- The Federal Office of Human Research Protections (OHRP)
- The study sponsor, the National Institutes of Health (NIH), including persons or organizations working with the sponsors

If a third-party gains access to your private health information, it may no longer be protected by federal regulations. **GINA** is a federal law that protects you from genetic discrimination by health insurance companies and employers. You are not protected under GINA for life, long-term care, or disability insurances.

Certificate of Confidentiality: To help protect your privacy, we received a Certificate of Confidentiality from the National Institutes of Health (NIH). With this Certificate, we cannot be forced to provide information that may identify you, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. We will use the Certificate to resist demands for information that identifies you, except as explained below.

The Certificate cannot be used to resist a demand for information from representatives of the US Government that is used for auditing or evaluation of federally funded projects or for information that must be provided in order to meet the requirements of the federal Food and Drug Administration. The Certificate does not stop you or a member of your family from telling others about yourself or your involvement in this research. If an insurer, employer, or other person gets your written consent to receive research information, then we cannot use the Certificate to withhold that information. If information regarding intent to harm yourself or others, including child abuse, becomes known to us, we are required by law to report this information even without your consent.

10. Future Use

After we remove your identifiable private information, your samples and/or data could be used for future research studies without additional consent from you. Researchers from other universities, the government, and drug-or health-related companies can apply to use the research materials. Your de-identified samples and data will only be shared with other researchers once they have gone through a review process and all of the appropriate regulatory and ethical approvals are in place. Any future testing or research using your samples and / or data may lead to the development and use of information, products, tests and/or treatments having commercial value. You will not receive any compensation that may result from these tests or treatments.

11. What is an Institutional Review Board (IRB)?

An IRB is a group of people that includes doctors, nurses, scientists, and community members. These people work together to make sure that research involving humans is well-placed and ethical. The IRB protects the rights and wellbeing of participants before and during the study. More information can be found on the Columbia University IRB website: http://www.cumc.columbia.edu/dept/irb

12. What if I change my mind?

You are allowing us to use and share your health information indefinitely However, you may choose to stop your participation in this study at any time and for any reason by emailing cpmg_info@cumc.columbia.edu or calling 212-851-4927. Your decision will not affect your care in any way. If you choose to stop your participation, the researchers may continue to use and share de-identified information they have already collected from you.

13. What if I have more questions?

If you have more questions about this project, contact the study team at cpmg_info@cumc.columbia.edu or 212-851-4927. For questions about your rights as a research subject, contact the CUIMC IRB at 212-305-5883 or askirb@columbia.edu. If you wish to receive genetic counseling prior to signing this form, we will help facilitate this at no cost to you.

STATEMENT OF CONSENT

I have read the consent form and talked about this research study, including the purpose, procedures, risks, benefits, and alternatives with the researcher. My questions were answered. I am aware that by signing below, I am agreeing to take part in this research study and that I can stop being in the study at any time. I am not giving up any of my legal rights by signing this consent form. I will be given a copy of the signed consent form and HIPAA authorization for my records.

Please **initial** the appropriate choices to show whether you give consent for the following items:

YES	NO		Initials
		I want to receive genetic results related to my current medical condition according to the procedure explained above.	
		I want to receive genetic results for additional medical conditions for which I may be at risk	
		I agree to be contacted in the future to have additional biological samples (like blood or saliva) collected and stored for this study.	
		I agree to be contacted in the future to inform me of other research opportunities	

Adult/13-17yo study participant and/or parent/legal guardian of participant

Print Name_____Signature_____Date_____

Print Name_____Signature_____Date_____

Print Name of the Child: _____

Relationship to the Child: _____

Person Obtaining Consent

Print Name:_____

Signature_____ Date: _____

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