

Columbia University Medical Center

Consent Form for Individual to Participate in a Genetic Research Study

[This template is intended to be used in studies for which genetic research is the focus. Options for storage and future use of biological samples and/or data for additional research objectives are included.]

This template may be used for (a) an individual with the medical condition for which sequencing or other genetic testing will be performed, (b) an individual without such medical condition (i.e., an unaffected family member or a healthy control), (c) the parent or legal guardian of a minor participant or (d) a legally authorized representative of a participant. There is a separate assent template for a child with or without such medical condition. For research involving trios (e.g., a child with such medical condition and his/her biological parents), a separate form should be signed for each participant. An adolescent who can understand this consent form may sign it in lieu of a separate assent form; in such cases, the parent/guardian should also sign this form to give permission for the minor to be enrolled.

This template does not apply to protocols that propose, as the primary aim: (1) the analysis of data from previously conducted sequencing or other genetic testing or (2) the establishment of a repository for collection of biological samples.]

Instructions for Consent Form Preparer:

Fill in the information requested in italics or delete as applicable. Include a version date in the footer. Please also ensure that a minimum 1" space is left in the lower right hand corner for the IRB approval stamp.

1. Title of research study and general information

Study title:

Study number: IRB-*[insert IRB protocol number]*

Participation duration:

2. Researchers' contact information

Principal Investigator:

Title:

Email address:

Phone Number:

Co-Investigator(s)/Study Coordinator(s): *[insert name(s) and title(s), as applicable]*

Email address(es):

Phone Number(s):

3. What information is on this form?

We are asking *[insert, as applicable: (a) you, (b) your child, (c) a minor for whom you are the parent or legal guardian or (d) the person for whom you are a legally authorized representative]* to take part in a research study.

[Include, if permission for participation may be provided by the research participant's parent, legal guardian or legally authorized representative:]

This consent form is written to address the research participant. If, however, you will be providing permission as *[insert, as applicable: (a) a parent, (b) a legal guardian of a minor or (c) a legally authorized representative]*, the words 'you' and 'your' should be read as *[insert, as applicable: (a) your child or (b) the research participant.]*

[Include, if applicable:]

A participant who is age 13-17 will *[insert, as applicable: (a) also be asked to read and sign this form or (b) will be asked to sign a separate assent form]* to indicate his/her willingness to participate in this study.

[Include, if applicable:]

A participant who is age 12 or under will also be asked to *[insert, as applicable: (a) read and sign an assent form or (b) provide verbal agreement]* to indicate his/her willingness to participate in this study.

This form explains why we are doing this study and what you will be asked to do if you choose to participate in it. It also describes the way we would like to use information about you and how we would like to use the *[select: (a) blood sample(s) and/or (b) tissue sample(s)]* ("biological samples") we obtain from you.

Please take the time to read this form. We will also talk with you about taking part in this research study.

If at any time you have questions about this form or the research study, please ask a member of the study team. Take all the time you need to decide whether you want to take part in the research study. Participation is voluntary; you do not have to participate if you do not want to.

4. Why is this study being done?

The purpose of this study is to gain a better understanding of the cause of *[describe the medical condition being studied]* (the "Study Medical Condition") through genetic testing. You have been asked to participate in the study because *[insert, as applicable: (a) you have the Study Medical Condition, (b) you are an unaffected family member of a subject who has the Study Medical Condition or (c) you are an individual whose genetic information will be compared to the genetic information of individuals who have the Study Medical Condition].*

We will study the results of the genetic tests being performed to find and possibly confirm associations between the Study Medical Condition and specific genes or genetic variants.

5. Some Background on Genes

We would like you to be well informed about genetic research, and for that reason we have, next, a few brief explanations. Please let us know, at any point, if you want or need more information in order to understand.

DNA is the material that governs the inheritance of many human traits, such as hair and eye color or the risk of some diseases. DNA is contained in most of the cells that make up the body's tissues. DNA carries the instructions for your body's development and functions.

A piece of DNA that determines a specific function of a cell is called a "gene." Abnormalities in the information in a gene can lead to disease.

Your entire unique genetic material, made up of DNA, is known as a "genome." An "exome" is the portion of the genome that includes only the DNA that is directly responsible for telling cells how to make the correct parts, or proteins, to function properly.

6. What are Whole Exome Sequencing and Whole Genome Sequencing?

We are requesting your permission to perform genetic testing on your biological samples to identify variants and consider their relationship to the Study Medical Condition. Genetic research is evolving rapidly. We expect that we will perform whole *[insert, as applicable: (a) genome and/or (b) exome]* sequencing but other genetic tests in addition to, or in place of, whole exome sequencing (WES) or whole genome sequencing (WGS) may be performed, including new genetic tests that may be developed in the future.

WES and WGS are very detailed types of genetic tests. WES searches through the exome for DNA variations that can cause disease. WGS searches through all of the genome, including areas outside of the exome. Because WES and WGS examine a larger portion of the genetic material than traditional tests, they may be able to find causes of disease where other tests did not. WES and WGS may also reveal information about unexpected diseases. Because WES and WGS are more comprehensive than other genetic tests, it is particularly important that you understand what is involved. *[Insert, as applicable: (a) The researcher conducting this study or (b) Your physician] [include, if applicable: and a genetic counselor]* will tell you more about WES and WGS. You may also wish to obtain professional genetic counseling prior to signing this consent form. *[Add a statement about whether genetic counseling is provided, and whether it is provided without cost or would be billable to the participant.]*

7. What will I be asked to do if I choose to be in this study?

If you agree to be in this study, we will schedule an evaluation with *[select, as applicable: (a) the researcher conducting this study or (b) your physician]*.

[If any of the following do not apply, remove them.]

Before this visit:

- We will collect some information about you, including your medical history.
- If you are a patient at Columbia University Medical Center (CUMC) and/or NewYork-Presbyterian Hospital (NYPH), we will review CUMC and/or NYPH electronic medical records and collect information, including *[insert specifics]*.
- If we require medical records from outside institutions, we will ask you to sign a separate authorization form to obtain them.

We will perform the following procedures during this visit *[modify as appropriate]*:

- Three-generation family history
- Detailed physical exam
- Photographs and video recording of the physical exam
- Blood draw of *[N]* tubes (*N* teaspoons) of blood
- *[Other]*

Genetic tests that are expected to include WES or WGS will be performed on the DNA in your biological samples.

[Add, if applicable:] We will also freeze some of the biological samples to repeat or confirm these tests or to perform further analysis for this study.

Sometimes we are unable to get sufficient DNA from the first set of biological samples that we collect, and may need to contact you to provide an additional sample so that the genetic tests can be conducted.

8. What results will be reported to me?

[Select one of the following alternatives:]

[Alternative No. 1, no plans to return results:] No results of the genetic testing will be reported to you as this is a research study.

[If Alternative No. 1 is selected, omit the remainder of Section 8. If Alternative No. 2 is selected, the remainder of this Section has an underlying premise that results may be returned.]

[Alternative No. 2, possibility of returning results:] If you are known to have the Study Medical Condition, results may be reported to you if the study team determines that a variant in your genome or exome is possibly, likely or definitely responsible for some or all features of the Study Medical Condition.

[If secondary findings may be returned, whether or not the subject has the Study Medical Condition include the following paragraph:]

Whether you have the Study Medical Condition or not, you may choose, at the end of this form, if you want to be provided with findings about conditions other than the Study Medical Condition that may be relevant to your health. These are generally called “secondary findings”. The nature of WES and WGS and other detailed genetic tests makes it possible that we may identify information about you that was not previously known, such as disease status or risk.

The study team may return genetic results to you if they determine that you have gene(s) or variant(s) that are probably or definitely associated with a medical condition. That condition may have been previously undiagnosed, or you may be at risk of developing it in the future. Knowing this information might help to prevent development of medical conditions. The absence of a reportable secondary finding does not mean that you have no disease-causing genetic changes, so if you have symptoms or features of a genetic disease in the future, clinical genetic testing should be considered. Coverage of specific genes through WES/WGS may not be as comprehensive as individual tests designed to investigate them.

[End of secondary findings section.]

[Include the following information if any results, primary or secondary, may be returned:]

Before any test results can be returned to you, they must be confirmed by a laboratory that is certified to provide clinical genetic testing. *[Add, if part of the initial sample was not stored in a CLIA-compliant environment for this purpose: You will be contacted to provide an additional sample for this testing.]* The purpose of additional testing in this laboratory is to confirm whether the variant is present. If the finding is confirmed, the results will be provided to your physician who will make a determination with the research team as to whether those results may have clinical importance to you. If they do, you will be notified and an appointment with your physician *[add, if applicable: and one or more members of the research team]* will be arranged to discuss the results.

If you are not known to have the Study Medical Condition, a positive test result can mean that you may be predisposed to (i.e., more likely to develop) or have the Study Medical Condition. If this is the case, you may wish to consider further independent testing, consult your physician or pursue genetic counseling.

If you are known to have the Study Medical Condition, it is important for you to understand that this study may not identify a cause for the Study Medical Condition because:

- The Study Medical Condition is not due to a genetic cause or
- A genetic change exists, but based on current knowledge, it cannot be determined whether it is related to the Study Medical Condition.

In these situations, you will be informed that the research analysis did not identify a genetic cause for the Study Medical Condition.

9. What about privacy and confidentiality?

Every effort will be made to keep your personal information confidential. However, we cannot guarantee total confidentiality.

[Describe the steps that will be taken to maintain confidentiality of subject data; see also Section 10:]

If information from this study is published or presented at scientific meetings, your name and other personal information will not be used.

The following people and/or agencies will be able to look at and copy your research records:

- The researchers, study staff and other professionals who are conducting the study or analyzing study results;
- If necessary for monitoring purposes:
 - Authorities from Columbia University, including the Institutional Review Board ('IRB'). An IRB is a committee organized to review, approve and oversee research involving human subjects.
 - The U.S. Office for Human Research Protections *[add, if applicable: and/or the U. S. Food and Drug Administration];*
- *[Add, if this study is sponsored.]* The sponsor of this study, *[add: sponsor name]*, including persons or organizations working with or owned by the sponsor may review your data for accuracy, but may not copy information with your name on it.

[If the study will use protected health information and a stand-alone HIPAA authorization form will be used, insert the following statement:]

You will be asked to sign a separate form to allow the use and disclosure of your protected health information.

[If HIPAA authorization will be combined with this consent form, add the authorization language here:]

10. What will happen to my biological samples and/or data?

As indicated above, if you agree to be in this study, you will provide biological samples that contain your DNA. The genetic tests will be performed on the DNA that is in these samples and such tests will produce genetic data about you.

Your samples and/or data may be retained for the life of this study. After the study is concluded, we will *[insert, as applicable: (a) destroy the samples and/or data or (b) with your permission, retain and use the samples and/or data indefinitely.]*

Whether or not your samples are destroyed at the end of the study,

[Select the applicable option:]

[If the research is funded by NIH, include:]

because our research study is funded by the U.S. National Institutes of Health (NIH), we are required to submit your genetic and/or clinical data in coded form to one or more databases managed by the NIH.

[If it is a requirement of participation in the study to allow the data of the participant to be included in another government or private database for research purposes, include:]

because our research study is funded by a sponsor that makes it a condition of participation to store your data, we will submit your genetic and/or clinical data in coded form to one or more *[if the research is funded by NIH, add "other"]* government or private databases developed to make data accessible to researchers. *[If the research is not funded by NIH, add "Some of these are managed by the National Institutes of Health."]*

[If storage of data is not required by the sponsor, add: We will request your permission later in this consent form to store and permit access to your data.]]

[If the data will be uploaded as unrestricted access data, include:]

Some of these databases permit public unrestricted access to the data.

[If the data will be uploaded as controlled access data, include:]

Some of these databases permit only controlled access to the data. Researchers who request access to data must promise that they will protect the data, only share data as permitted by the database rules, report any data breaches and not seek to identify any individual from the data.

[In either case, add:]

The data may be the combined or individual data of many people. Any data that is submitted will not be labeled with your name or other information that could be used to easily identify you. However, it is possible that the information from your genome, when combined with information from other public sources, could be used to identify you. We believe that this is unlikely to happen.

If you agree that we may retain and use your samples and/or data indefinitely, they will be stored at CUMC either with the researchers on this study or in a central storage facility called a repository in identifiable form in a coded manner. This means that 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 *[Select, as applicable: (a) on the researchers' or repository's data servers, (b) on an encrypted electronic device or (c) in a locked file cabinet.]*

Also with your permission, your samples and/or data may be used by other Columbia researchers or researchers at other institutions, including commercial companies, for research on the Study Medical Condition or other medical conditions. If they are given to researchers who are not researchers on this study, they will only be given in deidentified 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.

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 are the risks of participating in this study?

[If blood sample, select:] There may be slight pain or bruising due to the blood draw. We will use only skilled individuals to obtain blood from you.

[If tissue sample, insert relevant risk information:]

Even without your name and other identifiers, your genetic information is unique to you. There is a potential risk that someone will identify you from your genetic information or learn something about you by looking at your genetic information; this risk may increase in the future as technologies advance and more researchers study your genetic information.

The Genetic Information Non-discrimination Act is a federal law that prevents insurance companies from using your genetic information to deny health insurance coverage. The law also prevents employers from getting or using genetic information for employment-related decisions. However, the law does not prevent companies that provide life insurance, disability insurance or long-term care insurance from using genetic information.

[Include the following 2 paragraphs only if results may be returned:]

Current genetic testing is not an exact science, and you should be aware that the genetic testing being done in this study is considered research testing. As with all research, it is possible that although the test gives us information that we think may be important, we will not know what all of it means. Thus, it is possible that the meaning of the information you are given may change over time as additional research is conducted. The genetic research may identify genetic changes that may require additional testing to evaluate. This could result in anxiety, uncertainty and additional expenses that may or may not be covered by your insurance.

The genetic research may identify serious, untreatable genetic conditions. Such a finding can result in unexpected psychological trauma, both for you and your family. The detection of such a condition could also affect the health or health care needs of your siblings, children or other close relatives.

Because we cannot say with certainty how information derived from the genetic research could be used in the future, this study may involve risks that are currently unforeseeable.

12. Are there benefits to taking part in this study?

[Select the appropriate option, depending upon whether results will be returned]

[Alternative No. 1, for use if results will be returned:]

If you agree to take part in the study, and choose to receive results, there may be direct medical benefit to you. If a genetic predisposition for a medical condition is found, knowing this information may help

determine how to manage your medical care. We hope that in the future, information learned from this study will benefit other people with similar findings.

However, if the sequencing does not find information that would affect your medical care or well-being, there may not be any direct benefit to you. The knowledge gained may increase our understanding of genetic testing and results of genetic tests, and help patients in the future.

[Alternative No. 2, for use if results will NOT be returned:]

This study will not directly benefit you. However, information that is learned may increase our understanding of genetic testing and results of genetic tests, and help patients in the future.

13. Will I get paid or be given anything to take part in this study?

[Select:]

You will not receive any payment or other compensation for taking part in this study.

[Or, if there will be compensation to the participant:]

You will receive *[insert details of the compensation that will be provided]* for taking part in this study.

[Add, if total reimbursement within a calendar year will exceed \$600:]

According to U.S. Internal Revenue Service (IRS) regulations, compensation payments totaling more than \$600 in a calendar year must be reported to the IRS. We will need to obtain your Social Security Number for this purpose. Reimbursement for travel or other study-related expenses are not considered compensation for tax purposes.

[Add, if reimbursed for travel expenses:]

We will reimburse you up to \$ *[insert amount]* per visit for reasonable travel and parking expenses.

[Add, if reimbursement is by check:] You will need to provide the original receipt and your Social Security Number for reimbursement.

14. Will I incur costs if I take part in this study?

[Select:]

There will be no costs to you for being in this study.

[Or, if there will be costs to the participant:]

The study will pay for services that you receive because you are in the study such as *[list what will be paid for, e.g. blood tests]*. The study will not cover the costs of procedures or tests that you would have even if you were not in the study.

[Add, if results may be returned:]

You will not be charged for the costs of confirming in a clinical laboratory any findings to be used in research.

However, you or your insurance company will be responsible for any additional clinical test, including genetic tests that may be recommended by your physician as a result of information received from the study.

15. What are my rights if I take part in this study?

Taking part in this study is your choice. You can decide not to take part or stop being in the study at any time. Your choice will not involve any penalty or loss of benefits to which you are entitled and will not affect your access to health care at CUMC or NYPH.

You will need to notify in writing one of the researchers listed in Section 2 of this consent form if you decide to withdraw from the study before it is finished and no longer want to be contacted by the researchers.

You will need to specify in your written notice if you want your unused biological samples destroyed and your identifying information removed from all CUMC databases so that your samples and/or data will not be included in any future analyses. However, there are limitations on our ability to exclude your information or remove your biological samples after they have been de-linked from identifying information or deposited in scientific databases, and, if you have given your permission to do so, used or shared with other researchers.

16. Can I select someone to act for me in the future if I cannot act for myself?

You have the option to designate someone who can, if you lose the capacity to consent for yourself or die, make choices for you with respect to your data and/or biological samples.

17. Whom may I call if I have questions?

You may call *[insert name of Principal Investigator or study contact]* at telephone # *[insert phone number]* if you have any questions or concerns about this research study.

If you have any questions about your rights as a research participant, or if you have a concern about this study, you may contact the office below.

Human Research Protection Office
Institutional Review Board
Columbia University Medical Center
154 Haven Avenue, 1st Floor
New York, NY 10032
Telephone: (212) 305-5883
irboffice@columbia.edu

18. Why might researchers want to contact me in the future?

[Include this Section if permission for future contact will be requested.]

We may want to contact you for additional information or to get a new sample of your *[select (a) blood and/or (b) tissue]* in order to learn more about the research findings from this study. We may contact you directly or through your physician. We may ask you to provide a new sample or additional medical

information, participate in other research studies or allow us to use your samples and/or data in identifiable form for other studies. If a biological sample, your participation in future research or use of your samples in identifiable form is requested, you may be asked to sign an additional form to agree to this.

In addition, in the future, we may want to contact you if we learn more about the genetic basis for the Study Medical Condition or other medical conditions or if we are more certain about identifying the genetic cause of such conditions that might give you the opportunity to obtain treatment or better treatment for such conditions.

19. Selection of options (initials required for each decision)

Please initial your choice for each option. Your choices will not affect your status in this research study or your access to health care at CUMC or NYPH.

[Include only if results may be returned:] **Section 9: Reporting of Results:**

I agree **I do not agree** that you may notify me about the results of the genetic testing to be conducted as part of this study. *[Add, if laboratory consent for confirmatory testing will be obtained:]* I will be asked to sign a separate consent form for additional testing to confirm the results before information about them is given to me.

Section 16: Designation of a Proxy:

I do **I do not** wish to designate a proxy if I lose the capacity to consent for myself or die to: *[include option (a) only if results will be returned]* (a) receive the results of genetic testing that is done on my biological samples; (b) request that my biological samples no longer be used for this study; and/or (c) have the authority to have my biological samples transferred to another medical or research institution.

If applicable, please provide the name of the proxy: _____

Section 18: Storage and Future Use of Biological Samples and/or Data:

I agree **do not agree** to the **storage** of my samples and/or data at CUMC in identifiable form after completion of this study and the **use** of my samples and/or data in deidentified form for future research and/or testing, including for commercial purposes, that may or may not be related to this study.

You can change your mind regarding storage and future use of your samples, DNA and/or data at any time. Please see Section 15 of the consent form for further information.

Section 19: Future Contact

I agree] I do not agree] to being contacted in the future to provide an additional biological sample or medical information, to receive information about other research studies, with a request to use samples, DNA and/or data with my identifying information attached, or to receive additional information for the treatment of the Study Medical Condition or other medical conditions.

20. Statement of Consent

[When finalizing this document, please make sure the statement of consent and signatures are on the same page. If there are any large areas of blank space as a result, add the statement, "This section intentionally left blank."]

Statement of Consent

I have read this consent form and the research study has been explained to me. I agree to be in the research study described above.

A copy of this consent form will be provided to me after I sign it.

By signing this consent form, I have not given up any of the legal rights that I would have if I were not a participant in the study.

Signatures

[Omit signature lines that do not apply to your study. If the signature line remains, the expectation is that it will be used at the time of each enrollment or that "N/A" will be entered on the line at the time of enrollment.]

Research Participant _____ **Date** _____

Print Name of Research Participant _____

[If this consent also serves as the permission from a parent, legal guardian or legally authorized representative, please include the following signature line.]

Parent, Legal Guardian or Legally Authorized Representative _____ **Date** _____

Print name of Parent, Legal Guardian or Legally Authorized Representative _____

Person Obtaining Consent

Date

Print Name of Person Obtaining Consent

[Include a witness signature line if consent will be obtained from a Non-English Speaking individual using the short form process, or a person who is physically not able to read, talk or write.]

Witness

Date

Print name of Witness