

Columbia University Medical Center

Consent Form for Individuals with Medical Condition(s)/Symptom(s) to Participate in a Research Study Involving Whole Exome or Whole Genome Sequencing

[This template is intended to be used in studies for which whole exome and/or whole genome sequencing (WES/WGS) is the focus. An addendum includes options for storage and future use of biological samples and data after the sequencing is completed.]

*This template should be used only for an **individual with one or more medical condition(s)/symptom(s) for which sequencing will be performed** or the parent or legal guardian of a minor or a legally authorized representative of such individual. There is an additional consent template for an individual without a medical condition/symptom and an assent template for a child with or without a medical condition/symptom. Research involving trios (e.g., a child with one or more medical conditions and their biological parents) would utilize both of the applicable adult and parental consent forms.*

This template does not apply to protocols that propose, as the primary aim: 1) the analysis of data from previously conducted sequencing; or 2) 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. If your study has more than one consent form, clearly identify the individual forms 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: you, your child, the person for whom you are a legally authorized representative]* to take part in a research study.

[Include, if permission for participation is provided by the research participant's 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: the parent or legal guardian of a minor, a legally authorized representative]*, the words 'you' and 'your' should be read as *[insert, as applicable: 'your child' or 'the research participant.']*

[Include, if applicable:]

If your child is age 13-17, he/she will also be asked to read and sign this form to indicate his/her willingness to participate in this study.

[Include if applicable:]

If your child is age 12 or under, he/she will also be asked to *[insert, as applicable: read and sign a different form, 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 be in this study. It also describes the way we would like to use information about you and how we would like to use the *[select: blood sample(s), 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(s) or symptom(s) being studied]* through genetic testing. You have been asked to participate in the study because you have *[describe the medical condition(s) or symptom(s)]*.

We will study the results of the genetic tests being performed to find and possibly confirm associations between *[add the medical condition(s) or symptom(s) being studied]* and specific genes, genetic variants and/or genetic mutations.

5. Some Background on Genes

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 (WES) and Whole Genome Sequencing (WGS)?

We are requesting your permission to perform whole *[specify type of analysis, i.e., genome and/or exome]* sequencing on your biological sample(s) and link this to your *[select: medical condition(s), symptom(s)]*.

Whole exome sequencing (WES) and whole genome sequencing (WGS) are new 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 *[include, if applicable: and are experimental in nature]*, it is particularly important that you understand what is involved. *[Specify which doctor, e.g., The study, Your primary, Your treating]* doctor *[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 informed consent form. *[Add a statement about whether genetic counseling is provided without cost.]*

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: the study doctor, specify other doctor]*.

[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]*

WES or WGS will be performed on the DNA in your biological sample.

[Add, if applicable:] We will also freeze some of the sample for future examination to repeat or confirm these tests.

[Add, if applicable:] The sequencing and/or analysis of the sequencing data will be performed at the New York Genome Center. Biological samples, DNA and/or data that are sent to the New York Genome Center for this purpose will not contain information that can easily identify you.

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 sequencing can be conducted.

In this study, no tests other than those you have agreed to will be performed on your biological samples. We will *[select: destroy the samples after the testing is completed, retain the samples for no more than 60 days after they were collected (note that if you select either of the preceding two options, you must delete the section in the Addendum describing future storage and use), or request your permission to retain and use the samples indefinitely]*.

8. How are the results of WES or WGS interpreted?

Once the exome or genome is read, the information (“sequence”) that is obtained is analyzed for differences between your exome or genome sequence and a usual (or “reference”) sequence. Differences from the reference sequence are called “variants” or “mutations.”

If a particular gene is considered by the research team to be highly relevant to your medical condition(s) or symptom(s), this gene would be tested first. If testing is negative, some or all of the remaining genome or exome would be examined. *[If appropriate, revise to be applicable to this study.]*

The variations in your exome or genome will be compared with a list of mutations that are known to cause medical problems in other people with *[select: medical condition(s), symptom(s)]* similar to yours and are reported in the medical literature or in national or international databases. If a match is not found, *[select: the study team or other applicable person/group]* will then examine whether mutations

not previously described are present in genes that are known to cause the type of *[select: medical condition(s), symptom (s)]* you have.

9. What results will be reported to me?

Primary findings are results relating to the *[select: medical condition(s), symptom(s)]* being studied: *[Select one of the alternatives below for inclusion in this consent form.]*

[Alternative No. 1:] No results of the sequencing will be returned to you because this is a research study. *[If this option is selected, omit the remainder of this section, including Alternative No. 2 for primary findings, the secondary findings options and the return of results paragraph.]*

[Alternative No. 2:] Results may be returned to you if the study team determines that a variant present in your exome or genome is either (a) known to cause the *[select: medical condition(s), symptom(s)]* that you have or (b) may be important to your health, even if it has not been reported or seen before in people with your *[select: medical condition(s), symptom(s)]*.

Secondary findings are results not relating to the *[select: medical condition(s), symptom(s)]* being studied: *[Select one of the alternatives below for inclusion in this consent form.]*

[Alternative No. 1:] There are no plans to provide this information to you or your doctor because this is a research study. *[If Alternative No. 1 is selected, omit Alternative No. 2 for secondary findings, including the selection of options, from this section.]*

[Alternative No. 2:] You may choose, at the end of this form, whether or not to be provided with secondary findings. We will not be examining your exome or genome specifically for results not relating to your *[select: medical condition(s), symptom(s)]*. It is possible, however, that this study will identify information about you that was previously not known, such as disease status or risk. *[If applicable, add: We will examine your exome or genome for the genes that are on the list developed by [select: the American College of Medical Genetics and Genomics (ACMG), Baylor University, other source] of variations that are known to be associated with a disease or condition.]* The results might indicate that you have another previously undiagnosed, potentially serious condition or could develop it in the future. Some of these diseases might show up later during your lifetime and knowing about them might help to prevent development of medical conditions.

[End of secondary findings section]

[Include this paragraph if the study offers the option of return of results:] Before any test results can be returned to you, they must be confirmed. A DNA sample will be sent to 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 for an additional sample for this testing.]* This laboratory will confirm if the variant is present. If the finding is confirmed, the results will be provided to your *[specify which doctor, e.g. study, primary, treating]* doctor 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 *[specify which doctor, e.g., study, primary, treating]* doctor *[add, if applicable: and one or more members of the research team]* will be arranged to discuss the results.

A positive test result can mean that you may be predisposed to (i.e., more likely to develop) or have a medical condition that is being studied. You may wish to consider further independent testing, consult your physician or pursue genetic counseling.

It is important for you to understand that this study may not identify a cause for your *[select: medical condition(s), symptom(s)]* because:

- your *[select: medical condition(s), symptom(s)]* is not due to a genetic cause;
- a genetic change exists but, based on current knowledge, it cannot be determined whether it is related to your *[select: medical condition(s), symptom(s)]*.

In these situations, you will be informed that a genetic cause for your condition was not found.

10. 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; suggested procedures/text:]

All *[samples, DNA and/or data]* that are collected during the study will be stored in a **coded** manner. This means that your sample 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: on the researchers' data servers, on an encrypted electronic device, in a locked file cabinet]*.

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 may be 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 Office for Human Research Protections ('OHRP') *[add, if applicable: and/or the United States Food and Drug Administration ('FDA')]*;
- *[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 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].

11. Will my data be entered into a shared database?

[Select the applicable options, considering funding, plans to submit data for controlled or unrestricted access, and whether permission to upload data is a condition of participation.]

[If the research is funded by NIH:]

Because our research study is funded by the 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.

[For all research, include:]

[If the participant has the option to choose whether or not he/she wants his/her data to be included in a government or private database for research purposes, add "With your permission" at the beginning of the following sentence; omit if the participant does not have that option (i.e., it is a requirement of participation in the study to allow the data to be so included).] We may 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 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 data of many people or individual level data. 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 the sequencing and/or analysis of sequencing data will be performed at the New York Genome Center, include:]

The New York Genome Center may share data that cannot be linked to you with other researchers for any area of medical research, including research conducted at not-for-profit, commercial and academic research institutions.

12. 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 (GINA) 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 these 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. WES or WGS 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.

WES or WGS 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 WGS/WES could be used in the future, this study may involve risks that are currently unforeseeable.

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

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

[Alternative 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 cause for your *[select: medical condition(s), symptom(s)]* 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 new information, or if the information does not affect your medical care or well-being, there may not be any benefit to you directly. The knowledge gained may increase our understanding of genetic testing and results of genetic tests, and help patients in the future.

[Alternative 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.

14. 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 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.

15. 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 *[specify which doctor, e.g., primary, treating]* doctor as a result of the study.

16. 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.

If you decide to withdraw from the study, you will need to notify in writing the researchers listed in Section 2 of this consent form 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.

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

17. 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.

18. 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

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, Findings:**

☐ I agree ☐ I do not agree that you may notify me about primary findings.

[Include, if secondary results may be returned:]

☐ I agree ☐ I do not agree that you may notify me about secondary findings.

[Add, if applicable] I will be asked to sign a separate consent form for the testing to confirm the findings before information about them is given to me.

[Add, if secondary results may be provided and this option applies:] If you prefer, we can ask you at the time of notification of your primary test results whether or not you want to receive secondary findings information. If this is your choice, please indicate below.

☐ I agree that you may ask me at the time of notification of my primary test results whether or not I want to receive secondary information.

[Add only when the participant is being offered an option to allow this. If the research is NIH funded, this would not generally be an option unless the participant is also being asked for permission to allow uploading to a non-NIH funded database:]

Section 11, Data Sent to Database:

☐ I agree ☐ I do not agree to allow my data to be sent to one or more national *[add, if applicable: or international government, or private databases]* for use by other researchers. I understand that my data will be unable to be linked to me or will be coded with a unique code number for this purpose and the link to the code will not be shared outside of CUMC.

Section 17, 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 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: _____

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.]

Research Participant

Date

Print Name of Research Participant

[If this consent also serves as the permission from a surrogate, please include a "legally authorized representative" signature line.]

Legally Authorized Representative, Parent or Legal Guardian

Date

Print name of Legally Authorized Representative, Parent or Legal Guardian

Person Obtaining Consent

Date

Print Name of Person Obtaining Consent

Witness

Date

Print name of Witness

The signature of a witness is only required for minimal risk studies when obtaining consent from:

- a Non-English Speaking Research participant using the short form process, or
- a person who is physically not able to read, talk or write.