Individualized Medicine through the Application of Genetic Information

IRB-AAAQ4356 – Main study informed consent and HIPAA authorization form

Anticipated number of subjects: 500
Participation duration: Up to 14 months

Contacts

Contact	Title	Contact Type	Numbers
Wendy Chung	Associate Professor	Principal Investigator	Telephone: (212) 851-5313
Julia Wynn	Genetic Counselor	Genetic Counselor	Telephone: (212) 305-6987

Introduction

You are being invited to take part in a research study to determine the value of genomic screening in the general population, and specifically in Ashkenazi Jewish and Hispanic communities. We are interested in determining the extent to which people are interested in having genetic screening and how what they find out from their screening affects them and their healthcare. We will also be testing different ways of educating people about genetic screening to find the best way of communicating this information. The findings from this study will be used in planning a much larger study of genetic screening and in developing an approach to interpreting screening results. The purpose of this form is to give you information to help you decide if you want to take part in this research study. This consent form includes information about why the study is being done; the things that you will be asked to do if you are in the study; any known risks involved; any potential benefits; and options, other than taking part in this study, that you have.

In addition to this form, you should have received a link to a website or a DVD with more information about this study. If you decide to participate, you will have the opportunity to select how you want to learn about your results. You can select counseling sessions that are in-person, live through a video link, pre-recorded video, or by telephone.

This study is being done by a group of researchers at Columbia University Medical Center (CUMC), and is funded by Columbia University and the National Human Genome Research Institute. If at any time you have questions about the study, you can contact Aileen Espinal at 212 305-9314, Bianca Haser at 212 305-1573, or the research manager Julia Wynn at 212 305 6987. Take all the time you need to decide whether you want to take part in this research study.

Why is this study being done?

We are doing this research study because new advances in genetics allow physicians to look at a person's genes to learn whether that person is at increased risk for specific medical conditions in which genes play a key role. There are four types of information that are available through this testing. The four types are:

- whether a person is at increased risk to develop a serious medical condition that their doctors may be able to prevent, such as certain types of cancer or sudden cardiac arrest;
- whether a person has a greater chance of developing a medical condition that cannot be prevented based on current knowledge, such as a dementia like Alzheimer's disease, but that may affect planning for the later years;
- whether a person carries a gene that will not cause any problems for them, but could lead to problems for their current or future children or grandchildren.
- how a person's body breaks down and responds to certain medications and if a person may have a bad response to certain medications.

You are being asked to take part in this study because you received treatment at Columbia University Medical Center in the past two years, and you indicated when being screened for this study that at least two of your grandparents were Ashkenazi Jews or that you are of Hispanic/Latino descent.

Genomic research

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 in your body. DNA carries the instructions for how your body develops and functions. A piece of DNA that determines a specific function of a cell is called a "gene." Abnormalities in a gene can lead to disease.

We are requesting your permission to perform DNA sequencing on your biological samples to identify genetic factors that may relate to an increased risk of developing a genetic disorder or having a child with a genetic disorder. Genomic DNA sequencing examines your genes which are responsible for telling cells how to make the correct parts or proteins to function properly. Genetic factors are personal traits or characteristics that are inherited and run in families. The information obtained from these tests will include genetic information about you. To protect your identity, we will give your sample(s) a unique code number. Your name will not be linked to this code number. Because genomic sequencing is more comprehensive than other genetic tests, it is particularly important that you understand what is involved. You are also free to obtain professional genetic counseling prior to signing this informed consent form. You will be able to learn more about what you might be tested for by watching videos online or by speaking with a genetic counselor by phone or in person at no cost to you.

The Genetic Information Nondiscrimination Act (GINA) protects your access to health insurance. You cannot have your health insurance rates raised or denied because of any results from genetic testing. Theoretically, life insurance, disability, or long term care insurance companies could use information from genetic testing to deny coverage or raise the rate to applicants. Therefore, if you decide to participate in this research study and agree to receive genetic results, you may wish to consider the impact of these results on your insurability.

How are the results of genomic sequencing interpreted?

Once the DNA sequence is read, the information will be compared to the sequences of similar individuals to search for differences. Differences between your sequence and the other sequences are called “variants” or “mutations.” Your DNA sequence will be compared with a list of mutations that are known to cause the genetic conditions you choose to learn about. You will then be notified of the mutations we identify.

What results will be reported to me?

As part of this study, you will learn about a list of genetic conditions for which results could be returned to you. We will only let you know about mutations we find if they are among the genetic conditions for which you indicated you would like to receive results. All test results returned to you will be confirmed in a laboratory that is certified to provide clinical genetic testing. The results may be returned to you in different ways, depending both on your preferences and the results. Results for serious genetic conditions will be returned to you by a geneticist and a genetic counselor in a genetic counseling session that may conducted by telephone, video call, or in person, as you prefer. The meaning of any positive test results will be explained to you, and you will be provided with additional resources if necessary.

What is involved in participating in this study?

Additional information, questionnaire, and test kits

If you agree to participate, you will be asked to complete a questionnaire online or by pen and paper. This will include questions about your demographics, physical health, experience with disease, psychological status, and expectations for genetic screening. You do not have to answer any question that you find distressing or uncomfortable. The questionnaire should take approximately 30 minutes to complete.

You will then be given the opportunity to receive additional information about genetic screening through an in-person counseling session, live video counseling session, pre-recorded videos (online or via DVD), written materials, or telephone counseling session. Regardless of the pre-test counseling method chosen, everyone will be asked to complete a pre-test questionnaire to indicate their results preferences, what they thought about the pre-test education, and to answer a few questions before they proceed with genetic testing to ensure that they understand the kind of information that could be provided by these genetic screening tests.

Obtaining a blood sample and genomic screening

Once you have successfully completed the pre-test questionnaire, we will arrange to collect a blood sample for genomic screening. Once we have collected your blood sample, we will extract DNA and send it to a clinical lab to be screened for the conditions for which you elected to receive results. Depending on when you enroll for the study, your DNA sample may take up to twelve months to be screened for the genetic conditions you indicated. We will also freeze some of the sample for possible future use. However, the sample will not be used for subsequent studies without your consent.

Receiving findings from genomic screening

As indicated above, it may take one to 12 months for you to receive your genomic results. The laboratory will issue a clinical report once your results are ready. The clinical report will be sent to the genetic counselor at Columbia, who will provide you with the results you requested through your preferred method (in person, video

conference, telephone, mail, or online health portal). Any result that has personal health implications will be followed up with a telephone call offering a counseling session with a genetic counselor and clinical geneticist, by video or in person, regardless of how you elected to receive results.

Counseling will include discussion of the results and their implications, as well as limitations of the testing procedures. People whose results indicate an increased risk for a serious medical illness will, if they desire, be referred to other medical specialists or patient support groups. All participants whose genetic information is shared with them will be provided with a copy of their laboratory results and a letter summarizing the counseling session, including the implications of the genetic test results for future medical care, implications for other family members, and reproductive choices for the person and their family.

Adding your test results to your electronic health record

Regardless of your preferred method for receiving results, your results will be added to your electronic health record at the Columbia University Medical Center. This will make them available to all Columbia doctors. Additionally, the investigators will periodically look at your electronic health records for a year to see how your healthcare at Columbia University is affected by your genomic screening results.

Follow-up Questionnaires

You will be asked to complete additional questionnaires four weeks after you receive your results and eleven months after that. We will ask how you felt about receiving your genetic results and will assess your psychological well-being. We will also ask how you, your healthcare providers, and family members used the information, including its impact on the healthcare that you used, and about your feelings about having participated in genetic screening. We will also ask how comfortable you were with the way we presented information and whether you were satisfied with the method you chose for pre-test counseling and results disclosure. The questionnaires should each take about 30 minutes. You will be given a $25 Amazon gift card for completing the baseline and the two follow-up questionnaires, altogether three gift cards totaling $75.

Permission to audio record the genetic counseling sessions

We would like to audio record the genetic counseling sessions to study and better understand how participants experience these sessions and how to improve them. This recording will not be shared with anyone who is not involved in the study, and will be labeled with a study ID number and kept in a password-protected drive. Any mention of your name will be deleted from the file. You do not have to consent to be recorded in order to participate in the study.

Please indicate whether you consent to being audio recorded by checking the appropriate checkbox on the web form.

Permission to store your DNA and re-contact you

In the future, we may want to re-contact you for a number of reasons. Additional information may become available about your test results that we may offer to share with you. Such information may have implications for you or your family members, for example helping you or them make choices regarding preventive care or medical treatment. We may also want to contact you to see if you are interested in participation in other research. The benefits of allowing us to re-contact you include learning more about your test results and their implication for your or your family's health, and gaining new information from other research studies. The risks

are that some of the information that we offer you about health risks may be upsetting, but you will have the opportunity to decide whether you want to learn about new information. To have the opportunity to learn more about your test results and invite you to be part of other studies we would like your permission to store your DNA sample. Can we contact you in the future to participate in other studies and store your DNA for future use, including those that involve returning additional genetic information to you?

Please indicate whether you agree to storing your DNA for future use and being recontacted on the web form.

Will my data be entered into a shared database?

We will share your genetic information without any identifying information (i.e., de-identified) for research purposes that may benefit families with a variety of conditions. An example is a database called the database of Genotypes and Phenotypes (dbGaP) at the National Center for Biotechnology Information (NCBI). The genetic data submitted will be de-identified and the link back to any individual will only be retained by Columbia University under strict security. No one else will know your identity. The genetic and clinical information will be available indefinitely and will be accessible only to qualified researchers who will require NIH approval.

Permission to share your information with other studies

You may already be enrolled in other research studies that collect health information from you at Columbia University Medical Center. You can avoid having to provide the same information repeatedly by authorizing the sharing of your information between this study and other studies in which you are enrolled. You do not have to agree to share your information between this and other studies to participate.

Please indicate whether you consent to having your research information shared between studies on the web form.

Risks

There may be risks or discomforts if you take part in this study. The study may reveal information about your genes that could be upsetting to you. For example, it could show that you are likely to develop a serious medical condition that may or may not be preventable. Or it could show that you carry a gene that does not cause problems for you, but could lead to problems for your children. As a result, you could experience feelings of anxiety, sadness, depression, or guilt. To help you deal with the findings and the feelings they may lead to, you will have the option of speaking with a genetic counselor and clinical geneticist, and will be encouraged to do so. We can also refer you for additional counseling or mental health evaluation if needed. The costs of these additional services will not be covered by the study and will be billed to your health insurance as in standard care.

If the information from the genetic tests became available to other people or organizations, it could be used to discriminate against you. To reduce this risk, all results will be stored in a locked file or password protected database. All interviews and forms will be coded with a study number rather than your name, as will the blood samples that are collected from you. The code numbers and forms will be kept in separate locations. Each database will be password protected, with the password known only to the study coordinator, analyst, and Dr. Chung, the leader of this study. The results of your genomic screening will become part of your medical records. We will remind you of these issues again when the results are returned.

The Genetic Information Nondiscrimination Act (GINA) protects your access to health insurance. You cannot have your health insurance rates raised or denied because of any results from genetic testing. Theoretically, life insurance, disability, or long term care insurance companies could use information from genetic testing to deny coverage or raise the rate to applicants. Therefore, if you decide to participate in this research study and agree to receive genetic results, you may wish to consider the impact of these results on your insurability.

Benefits

You may or may not receive personal (direct) benefit from taking part in this study. You could learn that you or family members are likely to develop a particular disease, which could improve your doctors' ability to prevent the disease or to detect the disease earlier, when treatment is more likely to be helpful. In addition, you will receive genetic counseling concerning the risk of specific genetic diseases for you and for other family members. The benefits to society as a whole from this study include improving knowledge of how returning the information from genetic screening affects participants and help us to understand how best to do that.

What other options are there?

You may choose not to participate in this research study. Deciding not to participate will not affect your access to medical treatment at CUMC or your participation in any other research study.

What about confidentiality?

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

The data collected will be given a code number and separated from your name or any other information that could identify you. The research file that links your name to the code number will be kept in a password-protected database. Only the Principal Investigator and the study staff will be able to see this file.

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

Access to your health information is required to be part of this study. If you choose to take part in this study, you are giving us the authorization (i.e., your permission) to use the protected health information and information collected during the research that can identify you. The information we may collect and use for this research includes the following: Your name, address, telephone number, date of birth, email address, and certain demographic information; personal and family medical history from your medical records; genetic test results; and questionnaire responses.

The research information that is shared with people outside of Columbia University Medical Center and NewYork-Presbyterian Hospital will not include your name, address, telephone number or any other direct identifier unless disclosure of the information is required by law or you have authorized the disclosure.

The following people and/or agencies will be able to look at, copy, use and share your research information:

- The investigator, Columbia University Medical Center study staff, and other professionals who may be evaluating the study;
- Authorities from Columbia University and NewYork-Presbyterian Hospital, including the Institutional Review Board ("IRB"). An IRB is a committee organized to protect the rights and welfare of people involved in research.
- The Federal Office of Human Research Protections ("OHRP");

Your authorization to use and share your health information does not have an expiration (ending) date.

Once your health information has been disclosed to others outside of the Columbia University Medical Center, federal privacy laws may no longer protect it from further disclosure.

You may change your mind and revoke (take back) this consent and authorization at any time and for any reason. To revoke this consent and authorization, you must contact the Principal Investigator, Wendy Chung, MD, PhD, at 1150 Saint Nicholas Ave, Room 620, New York, NY 10032. However, even if you revoke this authorization, the researchers may continue to use and disclose the information already collected.

Certificate of confidentiality

To help us 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 any demands for information that would identify you, except as explained below.

The Certificate of Confidentiality 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.

The Certificate cannot be used to resist a demand from representatives of the U.S. Government for information that is used for auditing or evaluating projects that they are responsible for overseeing or for information that must be provided to meet the requirements of the federal Food and Drug Administration (FDA).

You should also know that this Certificate does not remove our responsibility to report certain communicable diseases, suspected child abuse, or danger of physical or mental harm to appropriate agencies or authorities.

What are the costs to me of being in this study?

There are no costs to you for taking part in this study.

Will I get compensated for being in this study?

Yes. If you join the study, you will receive a $25 Amazon gift card for completing each of the three questionnaires: the baseline questionnaire before you receive your results, and the two follow-up questionnaires after.

Do I have to be in this study?

Taking part in this study is your choice. You can decide not to take part in or stop being in the study at any time. Your choice will not affect the treatment you receive from doctors and staff at CUMC and New York Presbyterian Hospital.

You are free to drop out of the study at any time by calling Dr. Wendy Chung at (212) 851-5313. If you decide to drop out of the study, the information already collected from you will remain in the study database, identified only with your study number. If you want your information destroyed, you may request this. If you drop out, we will send you a letter indicating your wish to drop out or remove your information. We will allow ten days from the mailing of the letter to destroy the information just in case you change your mind. Once your information is destroyed, it cannot be used any further in this study.

What are my rights and responsibilities as a research subject?

- Being in the study is your choice.
- Your decision about being in the study or even dropping out will not change your care or healthcare benefits.
- By signing this document, you are agreeing to take part in the research study which includes the following: completing 3 questionnaires over the course of a year; donating a blood sample; allowing the collected sample to be used for genetic testing; having genetic counseling when the results are returned to you.

Whom do I call if I have questions or problems?

If you have any questions or concerns about the study, you may contact Dr. Wendy Chung at (212) 851-5313, Julia Wynn at (212) 305-6987, or Jennifer Kraszewski at (212) 305-5947.

If you have any questions about your rights as a research subject, you may contact the Institutional Review Board listed below.

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

Statement of consent and HIPAA authorization

Statement of consent and HIPAA authorization

I have read this consent and HIPAA authorization form. 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 and HIPAA authorization 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

Research participant

I consent to having the counseling session audio recorded as described on page four. ❒ Yes ❒ No

I consent to storing my DNA for future use and being re-contacted for future research studies, which may return additional genetic information to me. ❒ Yes ❒ No

I consent to having my research information shared between this and another studies at Columbia University ❒ Yes ❒ No

Print Name_____ Signature_____ Date_____