Columbia University Medical Center Consent Form

Attached to Protocol: IRB-AAAP9700
Principal Investigator: Wendy Chung (wkc15)
IRB Protocol Title: Congenital Anomalies Research Exploration

Consent Number: **CF-AAAS8526**
Participation Duration: one hour
Anticipated Number of Subjects: 1000

Contact

Contact	Title	Contact Type	Numbers
Priyanka Ahimaz	Certified Genetic Counselor	Study Coordinator	Telephone: 212-305-5091
Wendy Chung	Associate Professor	Principal Investigator	Telephone: 212-851-5313
Julia Wynn	Genetic Counselor	Study Coordinator	Telephone: 212-305-6987

Research Purpose

The purpose of the Congenital Anomaly Research Exploration (CARE) study is to examine why some people are born with birth defects. This study is a joint effort of many specialties who care for patients with birth defects including surgeons, pediatricians and geneticists. The goal of our study is to better understand the causes of birth defects to aid in treatment, diagnosis and prevention. When a suspect genetic cause is identified, we will establish a patient-specific cellular model of undiagnosed diseases by developing human induced pluripotent stem cells (iPSCs). Using these cells, we can better study the single cell physiology in these patients and develop more effective drugs. Our efforts will ultimately aid in the diagnosis, prevention, or treatment of diseases caused by rare and previously uncharacterized genetic variants.

This form is written to give you information that you can use to decide whether or not to allow your child to participate in the study

Information on Research

Introduction

The purpose of this form is to give you information to help you decide if you want your child to take part in a research study. This consent and HIPAA authorization form includes information about:

- why the study is being done;
- the things that your child will be asked to do if your child is in the study;
- any known risks involved;
- any potential benefit;

- options, other than taking part in this study, that you have; and
- the way your health information will be used and shared for research purposes.

The Research Coordinator will discuss the study with you. If at any time you have questions about the study, please ask a member of the study team. Take all the time you need to decide whether you want your child to take part in this research study.

The purpose of this research is described below in the 'What is Involved in This Study?' section of this consent form. This consent and HIPAA authorization form is written to address your child who is the research subject.

Description of genetic testing

We are requesting your permission to perform whole genome and/or exome sequencing on your child's biological sample(s) and link this to his/her medical condition(s). Whole exome sequencing (WES) searches through the exome for DNA variations that can cause disease. Whole genome sequencing (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. You can contact the principal investigator or one of the study coordinators if you wish to learn more about WES and WGS. You may also wish to obtain professional genetic counseling prior to signing this informed consent form.

What is Involved in this study?

Your child is invited to participate in a research study designed to help improve our understanding of the genetic causes of birth defects. Your child is invited to participate in this study because he/she is affected with a birth defect. If you decide to allow your child to participate, following the recommendation of your physician, a research coordinator will describe the procedures to be followed.

A) Study Questionnaire about your child's medical and family history
B) Blood sample with a maximum of 10ml (2 teaspoons) of blood taken by study personnel, OR
C) Saliva sample provided by your child into a collection kit
D) A skin biopsy collected at the time of a planned surgery or autopsy (optional)
E) Photographs of your child taken in your presence (optional)
F) A tissue sample, that would otherwise be discarded, collected at the time of a planned surgery (optional)
G) Any sample remaining from chorionic villus sample or amniocentesis that would otherwise be discarded (optional)
H) Developmental and behavioral assessments through questionnaire or by study personnel (optional)

The expected duration of your child's participation is one outpatient clinic visit lasting 30 minutes to one hour.

Tissue Samples

If your child has surgery or passes away, it may be helpful to analyze the tissue discarded from your child's surgery or tissue samples from your child's autopsy. We would only collect discarded tissue, or tissue that would otherwise be thrown away, so there would be no additional risk for your child other than the risk of the surgery. If tissue is collected from an autopsy, it would not affect the process or results of the autopsy.

_____(initial) I consent to the collection of discarded tissue and skin biopsy from my child's surgery or autopsy.

_____(initial) I do not consent to the collection of discarded tissue from my child's surgery or autopsy.

The tissue and/or blood obtained may either be retained by the researchers for medical, scientific, or educational research purposes or may be disposed of according to customary practices. If it is retained, it is possible that such research may result in the development of a product or process that is useful for diagnostic or therapeutic purposes. There are no plans to provide or share any financial gains that result from this research, which may have commercial value.

Cell Line

The investigator may wish to use a sample of your child's blood or tissue to create a cell line. A cell line, including human iPSCs, is a sample that is obtained and processed so that it can keep dividing in the lab and survive for a long period of time. This gives the investigator a constant source of genetic material (DNA) to be used for the purposes of the study. You may refuse to allow for the creation of a cell line, while still agreeing to have your child participate in the study.

_____(initial) I consent to the creation of a cell line.

_____(initial) I do not consent to the creation of a cell line.

Sample Storage and Future Research

We may want to use your child's blood and/or tissue sample(s) and clinical information in future research studies of birth defects as the methods for genetic analysis improve. If you agree let us keep your child's samples for future research, they may be kept forever. You have the right to have your child's unused samples or information kept about you for research purposes destroyed at any time. You can request this at any time by contacting Dr. Wendy Chung at Columbia University at 212-851-5313. Please initial below to indicate whether or not you give permission for us to store your child's samples.

_____(initial) I agree to have my child's specimens stored for research which is related to this study of my child's condition.

_____(initial) I do not agree to have my child's specimens stored for research which is related to this study of my child's condition.

Research Results

The research from your child's blood sample and/or additional testing may yield information that will immediately affect the health of your child or your family. It is possible that the researchers might find a genetic cause for the birth defect. In this case, the information can be reported to you or your child's physician. Please initial below to indicate whether or not you want to learn about research genetic test results.

_____(initial) I want to be contact with results.
_____(initial) I want my physician to be contacted with results.
_____(initial) I do not want to be contacted and I do not want my physician contacted with results.

Data Sharing

The genetic and clinical data generated through the research study will be de-identified and shared in one or more databases to be used for research purposes. An example is the NIH Database of Genotypes and Phenotypes (dbGaP). Access is granted only for research use. The genetic data submitted will be de-identified and only the principal investigator of the study will be able to link back the genetic data to the participant. No one else will know your child's identity. The sharing of these data may aid in the advancement of genetic research.

_____(initial) I consent sharing my child's de-identified genomic and clinical data.
_____(initial) I do not consent to sharing my child's de-identified genomic and clinical data.

Photographs

We will ask for pictures of your child's face (both frontal and profile) as well as any other features related to the birth defect which may include but is not limited to any physical birth defect outside of the face. The photographs may be used for educational purposes, lectures and publications. The images may be seen by members of the general public, in addition to scientists and medical researchers that regularly use these publications in their professional education. The photographs will not be published with any identifying information such as your child's name but it is still possible that someone may recognize your child.

The photographs may be entered in the DECIPHER website which is available to doctors (especially clinical geneticists) and scientists (especially cytogeneticists and molecular biologists). Any photographs posted on the DECIPHER database will be password protected and only available to clinicians/clinical cytogeneticists who are registered members of the DECIPHER consortium. Please initial below to indicate whether or not you give permission. The photographs will be kept for the duration of the study. By consenting to these medical photographs, you understand that neither you nor your child will receive payment from any party.

_____(initial) I consent to having my child's photograph used as noted above.
_____(initial) I do not consent to having my child's photograph used as noted above.

Future Contact

The Investigators may want to call me in the future to inquire about my child's health and/or to invite me or my child to participate in a follow-up study.

_____(Initial) I consent to future contact.
_____(Initial) I do not consent to future contact

Risks

When possible, the blood sample will be collected from an arterial line in place. If the blood cannot be collected from an arterial line, there small risk of bleeding at the venipuncture site and a small risk of developing a bruise at the site of needle insertion. This is a very small amount of blood, and donating this amount of blood will not pose any risks to your health. There is no additional risk associated with the collection of discarded tissue samples from surgery. The only risk is the expected risk of the surgery. There is no risk associated with the collection of tissue from an autopsy.
You should be aware that insurance companies sometimes use information on genetic testing to deny coverage to applicants. This study does not involve genetic testing; it is aimed at developing such testing for the future. Since that is the case, if you are asked, you should NOT report this as being genetic testing.

Benefits

Your child will not benefit from this study. Benefits to society may include further understanding of the genetic basis of disease. Ultimately this understanding can lead to significant improvements in the prevention and treatment of human disease.

Alternative Procedures

The alternative would be not to participate.

Compensation

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

Additional Costs

There will be no costs to you or payments made to you for your child's participation in this study.

Additional Information

Questions:

If you have any questions, please ask, and we will do our best to answer them. If you have additional questions in the future, you can reach the Study Coordinator at Columbia University Medical Center at 212-305-6987.If you have any questions on your rights as a research subject you can call the Institutional Review Board at 212-305-5883 for information.

Confidentiality

Any information collected during this study that can identify your child by name will be kept confidential. We will do everything we can to keep your data secure, however, complete confidentiality cannot be promised. Despite all of our efforts, unanticipated problems, such as a stolen computer may occur, although it is highly unlikely.

Access to your child's health information is required to be part of this study. If you choose to include your child 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 your child. The health information that we may collect and use for this research may include medical history that may be considered sensitive.

Information about your child may be obtained from any hospital, doctor, and other health care provider involved in your child's care that is needed for this research purpose, including information about the diagnosis and treatment of their birth defect.

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.

Biological samples and clinical data collected as part of this study will be coded with a study ID. The key linking your child's study ID to your identifying information is accessible only to the principal investigator and study coordinator.

Although every effort will be made to protect the confidentiality of your child's records, absolute confidentiality cannot be guaranteed. By signing this document you grant permission for information about your child obtained during the study to be made available to: The investigator, study staff and other health professionals who may be evaluating the study, Columbia University, New York Presbyterian Hospital, authorized representatives of the National Institutes of Health ('NIH'), Food and Drug Administration ('FDA'), the Office of Human Research Protections ('OHRP') or other

government regulatory agencies; and applicable Institutional Review Boards ('IRBs') that independently review the study to assure adequate protection of research participants, as required by federal regulations.

Once your health information has been disclosed to a third party, federal privacy laws may no longer protect it from further disclosure. Your authorization to use and share your health information does not have an expiration (ending) date. 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 at (212) 851 5313. However, if you revoke your consent and authorization, you will not be allowed to continue taking part in the research. Your decision not to participate in research will not affect your clinical care.

Voluntary Participation

Your child's participation in this study is completely voluntary. You can refuse to allow your child to participate or withdraw your child from the study at any time and such a decision will not affect your child's medical care at Columbia University Medical Center now or in the future. The investigator is also free to terminate the study, or your child's participation in it, at any time. You may at any time request that your child's blood and/or any other sample collected (or the materials in it, including genetic materials) be removed from our collection and destroyed. Signing this form does not waive any of your child"s legal rights.

Statement of consent and HIPAA Authorization

I have read the consent and HIPAA authorization form and talked about this research study, including the purpose, procedures, risks, benefits and alternatives with the researcher. Any questions I had were answered to my satisfaction. 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 waiving (giving up) any of my legal rights by signing this consent form. I will be given a copy of this consent and HIPAA authorization form to keep for my records.

I am the [ ] mother / [ ] father / [ ] legally authorized guardian of the child named below.

Signature

Parent/Guardian

Print Name_____Signature_____Date_____

Person Obtaining Consent

Print Name_____Signature_____Date_____

Subject Name

Print Name_____