Clinical Investigation Consent Form
The Rockefeller University Hospital
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You or your child are being asked to join a research study, which will take place at The Rockefeller University Hospital. This form tells about the research and hereafter “You” refers to you or your child. You should ask questions of the person who is explaining this form to you. After you feel that you understand the research, if you want to be part of the study, you will be asked to sign the form. You can always ask more questions and can later change your mind about staying in the study.

If you join the research study, you will take part for about 2 hours during 1 visit. The research study as a whole will last about 20 years.

About 20,000 people will take part in the research study.

Title of the research study:
The Genetics of Childhood Neurological Diseases

I. What this research study is about, and the reason for doing this research.

This study is being done to find out more about the inherited causes of neurologic diseases in childhood (genetic abnormalities of brain development such as mental retardation). We think that we can find differences in a person’s DNA that will explain the cause of their medical condition. The reason for doing this research is to identify these genes that cause or predispose to neurologic diseases. This will be important to help diagnose and treat the disease.

You have been asked to participate because you or a member of your family, possibly your child, has a neurologic disease thought to be caused by a genetic defect.

II. What is going to happen in this research study?

Consent Process: Informed consent is a process to help you understand the purpose of the research study, what will happen in the study, possible risks and benefits, and your right to withdraw from the study at any time. All of this information will be explained to you in detail. You should ask any questions you have until you feel that you understand what is asked of you to participate. You may then want to enroll, or you may decide not to join the
study. The decision to participate is entirely up to you. Even after the study has started, you may at any time ask more questions, or decide to withdraw from the study.

In this part, we explain the meaning of words that we are going to use to describe this study:

“Substances drawn from your body” refer to liquids such as blood or urine. It can also mean tissues such as skin, cells and DNA. Cells make up all parts of your body. DNA is inside all the cells of your body and carries your genetic or inherited information. When we draw blood, take tissue, or take other substances from your body, we are taking a “sample.”

“Cell line” means a group of cells that can live and grow outside of the body. They can also be frozen and can be used for future research.

A blood sample (approximately 3 tablespoons) will be drawn from a vein in your arm, or a saliva sample (approximately 2 teaspoons) will be collected from you. The amount of blood to be taken from your child will be appropriate to your child’s weight and not greater than 3 tablespoons. In order to identify the genetic cause of brain development in your family, we may also need to obtain a DNA sample of each member of your family that might also carry the diseased gene. This will be decided by Dr. Gleeson or one of his associates in advance, but may include your parents, children, siblings, cousins, nieces and nephews. Family members will only provide a blood sample if they consent to do so, and their refusal to participate will not impact the subject’s participation. DNA will not be obtained from members of your family not at risk for carrying the diseased gene.

We may ask you to provide a saliva sample either in person or via mail. If you are providing your sample via mail, we will send you a package containing an Oragene-DNA® salivary DNA self-collection kit and a postage-paid envelope. You will use the enclosed kit to provide your saliva sample. In order to provide the saliva sample, you will spit into the clear plastic tube until the collected saliva reaches the fill line (2 mL). You will then mail the sample, back to the researchers at the Rockefeller University using the enclosed postage-paid envelope.

After drawing your blood or saliva a sample of your DNA will be kept indefinitely and the researchers, their associates and successors will be responsible for deciding how it will be used. A small amount of your DNA will be sent to a DNA bank for Dr. Gleeson and other future researchers collaborating on the project to use. The sample will not include your name or any other identifying information but it will be sent with the name of the condition that we are studying in your family. Your sample may be used to validate new genetic mutations or to identify additional mutations in new genes involved in the condition in your family.

In this study you may be asked to have parts of your body photographed and/or videotaped. Your name will not be attached to any of your images and your identity will remain
confidential. These photographs and/or videotapes will not be shared with anyone outside the study team unless you sign a release form to have these shown at a professional conference or in a professional publication.

Some parts of this study are experimental. Here, the word “experimental” means that the test or the treatment is “not part of the usual routine care of patients”. This is a research study that searches for rare genetic changes in your DNA. While genetic tests for specific diseases have been available for many years, the testing methods that we are using are new and still regarded as experimental.

We will tell you or your doctor about any tests related to the research protocol that are performed by a New York State-approved laboratory, if the results may affect your health or safety.

By law, we cannot tell you or your doctor the results of experimental tests, that is, tests that have not been approved by New York State for diagnosis or treatment; however, if we find anything from experimental tests that might be important for your health, we may suggest that you have additional tests performed by a New York State-approved laboratory.

In this study, you will not receive routine care for any medical conditions related to this protocol.

In this study, you will not receive routine care for any other medical conditions you may have.

From time to time, we will make available to you a summary of interesting or general findings from this project describing how they contribute to our understanding of health and disease.”

Your medical information and test results will be written in your Hospital chart. The researchers may also keep separate records with information about you and your study tests.

Sometimes we will need to look at your earlier medical records. We will ask you to sign a form that will let health care providers share your records with us. This could be your doctor, a clinic or another hospital where you have been treated before.

III. What are the risks of taking part in this research study?

There may be some risks and discomforts in taking part in this study. We know that these risks and discomforts may happen during this study:

Potential side effects associated with having your blood drawn include discomfort, pain, bleeding, bruising, infection at the needle site, and fainting or feeling lightheaded.
Privacy Risks: There is the risk that there could be computer security breaches which could reveal your identity. There may be the risk that data about you may become public, and could be used by employers or law enforcement agencies. These privacy risks are described in greater detail below.

There may be other risks and discomforts that we do not know about now, but we will tell you any new information discovered which might affect your decision to participate or remain in the study.

IV. What are the alternatives to participating in this research study?

Your alternative to this study is not to participate

V. What are the benefits of taking part in this research study?

This study may be of no direct benefit to you or members of your family. Instead, others may benefit in the future from what we learn from this study

VI. Who will be able to see the information learned about you in this research study?

We will keep your personal information private, and will do our best to keep this information confidential. We will listen to what you say we may do with this information, and we will follow the law. For example, by New York State law, hospitals must inform the New York State Department of Health if we find that you have a reportable communicable disease, such as a sexually transmittable disease, like chlamydia, hepatitis, gonorrhea, syphilis and HIV-1. Also, the researchers must report to the authorities if they believe that child abuse or neglect has happened, or to prevent serious harm to you or others.

Whenever possible, data about you will be unlinked from your name and identified by a code. However, auditors and regulators from government agencies that oversee research, and people at the Rockefeller University Hospital and at Rockefeller University may see your information in the course of their duties.

Your privacy is very important to us and we will use many safety measures to protect your privacy. However, in spite of all of the safety measures that we will use, we cannot guarantee that your identity will never become known. Although your genetic information is unique to you, you do share some genetic information with your children, parents, brothers, sisters, and other blood relatives. Therefore, it is possible that genetic information from them could be used to help identify you. Similarly, it may be possible that genetic information from you could be used to help identify them. We may deposit your genetic data to databases/repositories available to others for research. While neither the public nor
the controlled-access databases/repositories developed for this project will contain information that is traditionally used to identify you, such as your name, address, telephone number, or social security number, people may develop ways in the future that would allow someone to link your genetic or medical information in a database/repository back to you. For example, someone could compare information in one database/repository with information from you (or a blood relative) in another database and be able to identify you (or your blood relative). It also is possible that there could be violations to the security of the computer systems used to store the codes linking your genetic and medical information to you. Since some genetic variations can help to predict the future health problems of you and your relatives, this information might be of interest to health providers, life insurance companies, and others. Patterns of genetic variation also can be used by law enforcement agencies to identify a person or his/her blood relatives. Therefore, your genetic information potentially could be used in ways that could cause you or your family distress, such as by revealing that you (or a blood relative) carry a genetic disease. If concerns over non-paternity or non-maternity arise, they will not be divulged under any circumstances.

There also may be other privacy risks that we have not foreseen.

If the researchers publish the results of this study, they will not mention your name or other information that could identify you.

**Genetic Information Nondiscrimination Act**

A Federal law, called the Genetic Information Nondiscrimination Act (GINA), generally makes it illegal in the United States of America for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. Additional information is available in the Outpatient or Inpatient Information Handbook.

**VII. What are the payment arrangements?**

There is no cost to you for being in this research study. You will not receive compensation for participating in this study.

If research using your samples helps develop a drug or another product that is sold to the public, the drug company, the University and the researcher may share in some of the profits. For example, a cell line from your samples could be used to make a product for sale. There are no plans to pay you any money resulting from such discoveries. However, by signing this form, you do not give up any rights you may have.

**VIII. What happens if you don’t want to stay in this study or your participation is ended?**

You can choose if you want or do not want to be part of this study. If you do not join, there
is no penalty and no one will hold this against you. If you decide to join this study, you may change your mind and stop taking part in the study at any time, and this will not be held against you. Information about you up to that time may stay a part of the study.

During this study, the researchers may learn new information that might make you change your mind about whether you want to stay in the study. You will be given that information promptly.

If you decide to join the study now but later want to stop, you should let the researcher know. If you decide later that you do not want the specimens collected from you to be used for future research, you may tell this to Dr. Gleeson, who will use his best efforts to stop any additional studies. However, in some cases, such as if your cells are grown up and are found to be generally useful it may be impossible to locate and stop such future research once the materials have been widely shared with other researchers.

The researchers also may stop you from taking part in this study, even if you do not choose to stop being in it. There are situations when, as a result of genetic testing, we can demonstrate that the father or mother is not in fact biological parent of the child. Non-paternity or non-maternity will not be revealed to the family, will not be reported in findings nor released to anyone else. In these rare cases we exclude the individual or a family in question from our study.

IX. Consent to the use, storage and sharing of your samples and data for separate research studies

The scientific value of your samples and the information obtained from them is greatly increased if we can share them with other scientists at universities and pharmaceutical companies worldwide. The genetic information obtained from your DNA is called genotype. The information about your disease condition and the physiology of your cells is called phenotype. May we:

- store, use, and share for many years your blood or tissue samples and data including genotype and phenotype data, with other investigators at Rockefeller and elsewhere, possibly worldwide, and including pharmaceutical companies, sample and/or data banks/repositories for separate studies for many years? Your samples will either be stripped of information identifying them as yours or coded (we will hold the key to the code) so that they cannot be identified as having come from you. Other data related to your sample, but that does not identify you may accompany the samples; and

- put anonymous data information from the analyses in a completely public database, available to anyone on the Internet; and

- put your coded genotype and phenotype medical data information and data information from more detailed analyses of your coded samples in a NIH controlled-access database/repository. The data information in this database/repository will be available only to qualified researchers from academic institutions and commercial organizations,
both domestic and foreign who have received approval from an NIH Data Access Committee?

Yes ______________  No ______________

Any time in the future, you may withdraw your consent to use any samples that have not already been used in research or shared. If you withdraw your consent, the remaining unused samples will be destroyed, unless the samples cannot be identified as having come from you. Data generated using your samples will continue to be used.

X. **Who do you call if a medical problem results from this research study?**

If you believe that this study has led to a medical problem, you should call the researcher listed below right away. The researcher will help you get appropriate, available medical care.

Name: Dr. Joseph Gleeson  
Phone No.: 212 327 8081 (business hours)  
Inpatient unit 212 327 8448 (after hours)  
Email: jogleeson@rockefeller.edu

The Rockefeller University does not plan to pay for medical care that you may have as a result of taking part in this study at The Rockefeller University Hospital. However, you do not give up any rights you may have to seek compensation by signing this form.

XI. **Who do you contact if you have questions about the research study?**

Please ask as many questions as you want about this research study and this consent form. If you agree to take part in this study and have questions later on, contact the following researcher:

Name: Dr. Joseph Gleeson  
Phone No.: 212 327 8081  
Email: jogleeson@rockefeller.edu

If you have any concerns about your experience while taking part in this research study, you may contact The Rockefeller University Institutional Review Board (IRB) Office at (212) 327-8410, or the Office of Clinical Research at (212) 327-8408.

XII. **May we have permission to contact you about future studies?**

May we contact you by phone to find out if you are interested in hearing about new research studies? Contact would be made by the Rockefeller staff of the Clinical Research Support Office for Recruitment. If you decide at any time that you no longer want to be contacted, please tell us, and we will stop calling you.

Would you like us to contact you about future research studies?
Yes ____________ No ____________
If you say “no” to this question, this will not affect your participation in this study.

AGREEMENT TO PARTICIPATE -- SIGNATURES REQUIRED

I have read this consent form, and my questions have been answered.

A copy of this consent form will be given to you. Please keep a copy of the form as it contains important information that you may wish to refer to during the research study and thereafter.

I hereby voluntarily consent to take part in this research study.

Name of the Study Participant (Print) __________________________________________________________

_______________________________________________________________________________________

Signature of Study Participant Date (To Be Filled in by Study Participant)

_______________________________________________________________________________________

ALTERNATE SIGNATURE BLOCK

Participant requires assistance by a translator
Translation Services Provided by (choose one, by checking one box below):

☐ Pacific Interpreters

_______________________________________________________________________________________

Language Translator Identification Number

Witness to telephone translation: ________________________________ (Print Name)

_______________________________________________________________________________________

Signature of witness Date

☐ Other Translator:

_______________________________________________________________________________________

Name of translator Date

Witness to oral presentation: _________________________________________________________________
ALTERNATE SIGNATURE BLOCK

Adult not legally capable of giving consent

Name of the Study Participant (Print) ________________________________

Name of Legal Representative (Print) ________________________________

Signature of Legal Representative Date (To Be Filled in by Representative)

I hereby voluntarily consent to have my child take part in this research study.

Name of Child (Print) ________________________________

Name of Mother or Legal Guardian (Print) ________________________________

Signature of the Mother or legal guardian Date (To Be Filled in by Mother or Guardian)

Name of Father or Guardian (Print) ________________________________

Signature of the Father or legal guardian Date (To Be Filled in by Father or Guardian)

ALTERNATE SIGNATURE BLOCK
Signature of the Person Conducting the Informed Consent Discussion

I have explained the research protocol and this consent form to the participant and have answered the participant’s questions about this research study and/or the consent process.

Name of Person (Print) ________________________________________________

____________________________________  ______________________________
Signature of Person Discussing Date (To Be Filled in by Person Discussing
Consent) Consent

Rockefeller University Institutional Review Board
IRB NUMBER: JGN-0853
IRB APPROVAL DATE: 06/12/2014
IRB EXPIRATION DATE: 06/04/2015