

Subject's Name: \_\_\_\_\_  
Title of Research Protocol: The Genetics of Childhood Neurological Diseases  
Investigator's Name: Dr. Joseph Gleeson  
Protocol Number and expiration date: 140028 At the completion of this study

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**University of California - San Diego  
Adolescent Assent Form**

Joseph G. Gleeson, MD and his associates are conducting a research study to find out more why some children have brain diseases such as seizures and mental retardation. This research project studies the genes from children with brain diseases and their parents, in order to pinpoint which genes are responsible for brain diseases. You are asked to participate because someone in your family either has mental retardation or convulsions or may be at risk for developing these sorts of problems. There will be approximately 800 new participants joining this study every year. To date there have been over 10,000 individuals participating in this project.

If you agree to be in this study, the following will happen to you:

1. A blood sample (approximately 3 tablespoons) will be drawn from a vein in your arm, or if you are having surgery, from the IV line that is placed for anesthesia, or a saliva sample (approximately 2 tablespoons) will be collected from you and sent to Dr. Gleeson's laboratory.
2. If you are having surgery, the surgeon will save a small piece of tissue that would have been thrown away otherwise.
3. If you have an HPDL mutation, we may request an additional blood draw (about half a teaspoon) to analyze metabolome levels. We may also ask if you agree to undergo neurological examinations at a clinic, neurodevelopmental tests, and various assessments of neurological functions. Additionally, we might request you to complete questionnaires and interviews that assess various aspects of neurologic function, adaptive behavior, and quality of life for yourself and caregivers. In total, assessments can take between 1-3 hours per appointment.
4. Medical records from each member of your family may be forwarded to Dr. Gleeson for review.
5. After drawing your blood or saliva, and surgical tissue if applicable, Dr. Gleeson will be responsible for deciding how it will be used. The sample may be used for additional research carried out in Dr. Gleeson's lab. Your sample can be shared with current and future researchers collaborating on this project. The sample will not include your name or any other identifying information, but it will be sent with the name of the disease that we are studying in your family. Once identifiers have been removed, we will not ask for your consent for the use or sharing of your specimens in other research. Your sample may be used to validate new genetic mutations or to identify additional mutations in new genes. You consent to such uses. If you later decide you do not want your blood used for research, you can tell Dr. Gleeson and we will do our best to destroy your blood sample.
6. Dr. Gleeson will be responsible for deciding how your sample will be used. The sample collected may also be used in additional research to be conducted by the University of California personnel collaborating in this research. These samples and their derivatives may have significant therapeutic or commercial value. You will not

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share in any commercial value or profit derived from the use of your biospecimens and/or information obtained from them. You consent to such uses.

7. You will not receive compensation for participating in this study, though you may be reimbursed for your travel expenses. Additionally, the necessary phlebotomy procedures or doctor visits will be of no cost to you.

Participation in this study may involve some added risks and discomforts. These include:

1. The blood draw may hurt slightly. Risks include possible pain, discomfort, and bruising at the needle site, possible dizziness and fainting and possible infection. Any associated bleeding will be treated with pressure and the area may bruise temporarily. If infection occurs, it will require treatment.
2. There is a chance that participation in this study could cause psychological distress. Some people involved in genetic studies have felt anxious about the possibility of having a gene that places them at risk or that can be passed on to children. If you experience these feelings during the study, have your parents contact us and we will arrange for you to speak with a genetic counselor.
3. Risks of Genetic Testing: Federal and State laws generally protect your genetic information in the following ways:
  - a) Health insurance companies and group health plans may not request your genetic information from this research.
  - b) Health insurance companies and group health plans may not use your genetic information when making decisions regarding your eligibility or premiums.
  - c) Employers with 5 or more employees may not use your genetic information from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment.Be aware that these laws **do not** protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. We will minimize the possibility of results from this research being linked to you, but there is always the remote possibility that information from the research may be disclosed. If your genetic risk for certain diseases is accidentally divulged to the wrong source, you might be discriminated against in obtaining life or health insurance, or employment.
4. We may need to review your medical records such as MRI scans, pathology reports, laboratory or progress reports etc. This information as well, will be kept confidential and not shared with anyone outside this project. We will not release this information to insurance companies, family members, workplaces or any other institutions. Even though, the risk of losing confidentiality via medical records cannot be fully prevented.
5. In order to help advance future patient screening strategies, a new condition in our research study might require that your DNA sequence, diagnosis, and de-identified pedigree data be deposited into the NIH's (National Institute of Health) dbGAP or similar databases. Your personally identifiable information (PII) will not be shared on this database. Your PII will remain confidential. Only de-identified genetic data would be deposited into the dbGaP database.

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While the public database will not contain information traditionally used to identify you, people may develop ways in the future that would allow someone to link your genetic information in these databases back to you. For example, someone could compare information in a database 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 information to you.

While your privacy and confidentiality are very important to us and we will use safety measures to protect it, we cannot guarantee that your identity will never become known.

Dr. Gleeson and/or \_\_\_\_\_ have told you about the study and have answered all your questions.

Your other option is not to participate in this study. You do not have to be in this study if you don't want to be. You can stop any time you want and no one will be upset or angry with you.

You agree to be in this study:

\_\_\_\_\_  
Adolescent's Signature

\_\_\_\_\_  
Date

\_\_\_\_\_  
Witness

\_\_\_\_\_  
Date