University of California - San Diego
Adult Consent Form

Joseph G. Gleeson, MD and his associates are conducting a research study to find out more about the inherited causes of brain diseases in childhood (genetic abnormalities of brain development such as mental retardation). You have been asked to participate because a member of your family, possibly your child, has a neurological disease thought to be due to a genetic defect. The goal of this study is to identify the specific genetic defect underlying the condition in your family. 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. In order to localize the genetic cause of brain development in your family, we also need to obtain DNA sample from each member of your family that might also carry the diseased gene. This determination will be made by Dr. Gleeson or one of his associates in advance, but may include your parents, children, siblings, cousins and nieces/nephews. DNA will not be obtained from members of your family not at risk for carrying the diseased gene.
4. You may be asked to sign a release to allow your medical records to be forwarded to Dr. Gleeson for review.
5. If you choose, you will be notified of results obtained through this study. We will not disclose non-maternity or non-paternity information.
6. 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 of drawing blood include possible pain, discomfort, and bruising at the puncture site, possible dizziness and fainting and possible infection. Prolonged bleeding is treated with pressure to the needle site, and bruising may leave the needle site temporarily discolored. If infection occurs, it will require medical attention.
2. There is a chance that participation in this study could cause psychological distress, economic and social harm. Some people involved in genetic studies have felt anxious about the possibility of carrying an altered gene that places them at risk or that can be passed on to children. If these feelings arise at any time during the study, you may 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 accidently 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. We will not release this information to insurance companies, family members, workplaces or any other institutions. Even though, the risk of loosing 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 database. 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 or similar databases.
   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.
6. Non-paternity or non-maternity refers to the situations when as a result of genetic testing we are able to indicate that reported relationship between one or both parents with one or multiple children is inconsistent with the genetic make-up. In other words, based on the DNA analysis, 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.

What will happen to information and/or biospecimens collected from me?
After drawing your blood, or taking saliva or surgical tissue if applicable, a sample of your DNA will be kept with Dr. Gleeson indefinitely, and Dr. Gleeson, his associates or successors in these studies will be responsible for deciding how it will be used. In addition to Dr. Gleeson, your genetic material 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 condition 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 involved in the condition in your family.

The specimens collected from you and the DNA that they contain may also be used in additional research to be conducted by the University of California personnel collaborating in this research. These specimens, DNA, and their derivatives may have significant therapeutic or commercial value. You will not 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.

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 at the University of California.

If you are injured as a direct result of participation in this research, the University of California will provide any medical care you need to treat those injuries. The University will not provide any other form of compensation to you if you are injured. You may call the Office of IRB Administration at (858) 246-4777 for more information about this, to inquire about your rights as a research subject, or to report research-related problems.

This study may be of no direct benefit to you or members of your family. If as a result of participation in this study we obtain information that could significantly affect your health or well being, we will attempt to inform you of the existence of this information. You may then decide if you wish to know what we have learned. Dr. Gleeson hopes to be able to identify the gene responsible for the condition in your family, as well as develop improved diagnostic procedures and possibly new methods of treatment. In addition, this information may also be used to further our understanding of neurological disorders in other individuals and families.

Instances are known in which a subject in a research study has been required to furnish genetic information as a precondition in applying for health insurance and/or a job. Participation in this study does not mean that you have had genetic testing. Genetic testing means having a test performed and the results provided to you and your doctor. If you are interested in having genetic testing performed, you should consult your doctor, as some commercial tests are available. Your doctor can provide you with the necessary information to determine if such a test would be appropriate for you.

Dr. Gleeson and/or _____ on Dr. Gleeson's behalf has explained this study to you and answered your questions. If you have other questions or research-related problems, you may reach Dr. Gleeson at (858) 246-0547.

Your alternative option to participation in this study is not to participate. Your involvement in research is entirely voluntary. You may refuse to participate or withdraw at any time without jeopardy to the medical care you will receive at this or any institution.
Research records will be kept confidential to the extent provided by law.

You have received a copy of this consent document and a copy of "Experimental Subject's Bill of Rights" to keep.

You agree to participate.

_____	_____	_____
Subject's Signature	Witness	Date