**Rady Children's Hospital – San Diego**
**and University of California, San Diego**

**Parental Informed Consent**

**The Genetics of Childhood Neurological Diseases**

This is a research study. Research studies include only subjects who choose to take part. You are being asked to let your child take part in this study because your child has a neurological disease thought to be due to a genetic defect. Please take your time to make your decision. Discuss it with your child and family. Be sure to ask any questions that you may have.

## STUDY INVESTIGATOR

Investigator(s): Dr. Joseph G. Gleeson, MD

## WHY IS THIS STUDY BEING DONE?

The goal of this study is to identify the specific genetic defect underlying the condition in your family.

## HOW MANY PEOPLE WILL TAKE PART IN THE STUDY?

Approximately 800 subjects will be enrolled in this study each year, including around 30 at RCHSD, 50 nationwide (including RCHSD) and 750 internationally.

## HOW LONG WILL YOUR CHILD BE IN THE STUDY?

Your child will be in the study until it expires.

You can stop your child's participating at any time. However, if you decide to stop your child from participating in the study, we encourage you to talk to the research doctor.

## WHAT IS INVOLVED IN THE STUDY?

This is what will happen if your child participates in this study:

1. A blood sample (approximately 3 tablespoons) may be drawn from a vein in your child's arm or IV line if during surgery, or a saliva sample (approximately 2 tablespoons) will be collected from your child and sent to Dr. Gleeson's laboratory.
2. If your child is having surgery, the surgeon will save a small piece of tissue that would have been thrown away otherwise.
3. If your child has 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 have your child undergo neurological examinations at the 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 both your child and yourself as caregivers. In total, assessments can take between 1-3 hours per appointment.
4. In order to localize the genetic cause of brain development in your family, we may also need to obtain a DNA sample in the form of saliva or a dried blood spot of 3-5 drops of blood obtained through a finger prick from other family members.
5. You may be asked to sign a release to allow your child's medical records to be forwarded to Dr. Gleeson for review.
6. If you choose, you will be notified of results obtained through this study. We will not disclose non-maternity or non-paternity information.
7. After drawing your child's blood, taking a dried blood spot, or taking saliva, 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. 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 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 sample.
8. 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 and 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.
   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.

## WHAT ARE THE RISKS OF THE STUDY?

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

1. The blood draw or finger prick in case of dried blood spot may hurt your child 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.

For more information about these risks and side effects, ask your child's study doctor.

## ARE THERE BENEFITS TO TAKING PART IN THE STUDY?

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.

## CAN YOUR CHILD BE REMOVED FROM THE STUDY WITHOUT YOUR CONSENT?

Yes. If information in your medical records indicates that your child's condition is not amenable to genetic study, they may be removed from the study.

## WHAT ABOUT CONFIDENTIALITY?

Every reasonable effort will be made to keep your records confidential. All of the information we collect during the study will be kept locked and secured. None of it will be released unless your parents ask us to do so. However, while you are in this study we do have to let some people look at your records. These people can see your records:

- The UCSD Institutional Review Board (for the protection of human subjects in research)
- Gleeson lab members and collaborators working in this research project.

We will keep your records confidential unless we are required by law to share any information.

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, work places or any other institutions. Even though the risk of losing confidentiality via medical records cannot be fully eradicated, we take all the precautions to protect this information.

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 the study results are published or presented, your child will not be identified.

## WHAT ARE THE COSTS?

The necessary phlebotomy procedures or doctor visits will be at no cost to you.

## WHAT IF YOUR CHILD IS INJURED IN THE STUDY?

If your child is injured as a direct result of participation in this research, Rady Children's Hospital – San Diego or the University of California will provide any medical care needed to treat those injuries. Neither Rady Children's Hospital – San Diego nor the University will

provide any other form of compensation to you if your child is injured. You may call the Office of IRB Administration Office at (858) 246-4777 for more information about this, to inquire about your child's rights as a research subject or to report research-related problems.

## WILL YOU OR YOUR CHILD BE COMPENSATED?

You will not receive compensation for participating in this study, though you may be reimbursed for your travel expenses.

## WHO DO YOU CALL IF YOU OR YOUR CHILD HAVE QUESTIONS OR PROBLEMS?

For questions about the study or a research-related injury, contact the researcher:

**Dr. Joseph Gleeson (858) 246-0547**

## WHAT ARE YOUR CHILD'S RIGHTS AS A RESEARCH SUBJECT?

Taking part in this study is voluntary. You may choose not to let your child take part or you or your child may choose to leave the study at any time. Your decision will not result in any penalty or loss of benefits to which your child is entitled. If you have questions about your child's rights you may call:

University of California, San Diego
Office of IRB Administration
(858) 246-4777

You will be told about any new information that may affect your child's health, welfare, or willingness to stay in this study.

## SIGNATURE AND CONSENT TO BE IN THE STUDY:

Your signature below means that you have read the above information about the Genetics of Childhood Neurological Diseases study and have had a chance to ask questions to help you understand what your child will do in this study and how your child's information will be used.

You or your child can change your minds later if you want to. You will be given a copy of this consent form and a copy of the Subject's Bill of Rights. By signing this consent form you are not giving up any of your or your child's legal rights.

You agree to allow your child to participate in this research study.

_____ _____

NAME OF PARTICIPANT AGE

_____ _____

SIGNATURE OF PARENT OR GUARDIAN DATE

_____ _____

SIGNATURE OF WITNESS (person explaining this form) DATE

## SUBJECT'S BILL OF RIGHTS

It is important that the purpose and procedures of the research study are fully understood and that consent is offered willingly. A subject in a research study or someone, who is asked to give consent on behalf of another person for such participation, has the right to the following:

1. Be informed of the nature and purpose of the research.

2. Be given an explanation of all procedures to be followed and of any drug or device to be used.

3. Be given a description of any risks or discomforts, which can be reasonably expected to result from this research study.

4. Be given an explanation of any benefits, which can be reasonably expected to the subject as a result of this research study.

5. Be informed of any appropriate alternative procedures, drugs, or devices that may be advantageous and of their relative risks and discomforts.

6. Be informed of any medical treatment, which will be made available to the subject if complications should arise from this research.

7. Be given an opportunity and encouraged to ask any questions concerning the study or the procedures involved in this research.

8. Be made aware that consent to participate in the research may be withdrawn and that participation may be discontinued at any time without affecting continuity or quality of medical care.

9. Be given a copy of the signed and dated written consent form.

10. Not be subjected to any element of force, fraud, deceit, duress, coercion, or any influence in reaching the decision to consent or to not consent to participate in the research.

If you have any further questions or concerns about your child's rights as a research subject, please contact your research doctor or the UCSD Office of IRB Administration at (858) 246-4777.