Boston Children's Hospital

RESEARCH CONSENT FORM

DO NOT PLACE IN MEDICAL RECORD

Use Plate or Print:

Subject Name:

DOB:

Gender:

Protocol Title: Genetics of Epilepsy and Cognitive Disorders

Principal Investigator: Christopher Walsh

Description and Explanation:

The purpose of this research study is to investigate the genetic causes of disorders of human epilepsy and cognition. These include disorders of brain formation such as microcephaly (small brain), lissencephaly (smooth brain), and polymicrogyria (too many small folds in the brain surface), disorders of brain function such as mental retardation and autism, and other related conditions. You have been asked to be a part of this study because you or a member of your family has been diagnosed with one of these disorders. This consent form gives you detailed information about the procedures, risks and benefits of the study so that you can decide whether or not you want to be a part of this research.

Genes are found in the cells of our body and are the instructions that tell our body how to grow and develop. They are passed on to us (inherited) from our parents and most genes are the same in each of us. There are harmless changes in our genes that make each of us unique, such as in genes causing eye color and height. There can also be changes in genes that cause them not to work properly and lead to health problems or disease. We wish to determine and understand which genes are important for brain development by studying the changes in genes of individuals who have disorders of brain development. We hope that the knowledge we gain from this research will lead to improved diagnosis, management and treatment of these conditions.

Subject Selection:

You have been asked to be a part of this study because you or a member of your family has a disorder of brain development as described above. Since 2002, individuals and their family members from Boston Children's Hospital, across the United States and around the world have been enrolled in this study. We hope to include over 2000 families in this research in order to identify the many genes important for brain development.

Procedure:

Interview: If you choose to participate, you will be interviewed by telephone or in person about the medical history for you, your child and your family members. Questions will focus on the pattern and development of the disorder of brain development in your family. Other questions may include age, ethnic background, and biological relationships between individuals. We may review your hospital records or contact your health care provider. You may also have a brief neurological exam that could include testing your coordination, muscles, memory, and/or language and concentration skills. This may be done by your own doctor or by a study doctor at Boston Children's Hospital and should take less than one hour. You will not be charged for this exam.

Sample Collection: You will be asked to give a blood sample, from which we can study your genetic material (genes and DNA). We request that a blood sample of up to 4 teaspoons (20cc) be taken from a vein in your arm using a sterile needle. This should only take a few minutes and would be 5cc (1 teaspoon) of blood from children under 2 years, 5-10cc (1-2 teaspoons) from children 2-8 years old and 10-20cc (2-4 teaspoons) from children over 8 years and adults. If there are costs for your blood draw or transportation to the appointment to have your blood drawn for participation in this research, we will refund these costs to you if you give us the receipt(s) showing the exact cost and date of service.

For some individuals we may ask for a different sample to be able to obtain your genetic material. Other samples could be cheek cells from a small brush used on the inside of your cheek, a saliva sample from spitting into a small container, a urine sample collected in a sterile container, or a hair sample from pulling a few hairs from your head. If you had a surgical procedure in the past and a tissue sample is saved that is not needed for other purposes, we may request some of this tissue.

Magnetic Resonance Imaging (MRI): If you have a disorder of brain development, we may suggest that your doctor perform an MRI of your brain if it is clinically indicated. An MRI is often the standard care given to patients with epilepsy and could provide the best description of your condition. If a satisfactory brain MRI has not been done in the past, we may invite you to have an MRI at our expense so that the best possible diagnosis is made. For the MRI, you would need to lie still for about 45 minutes while pictures of your brain are taken. A computer puts these pictures together and the results are usually available a few days later. At your request, a copy of the MRI report would be sent to you and your doctor and you could receive a copy also.

Research Uses of Samples:

We will use the genetic material from your sample(s) to identify and study genes involved in disorders of brain development. This could involve different types of laboratory techniques used for examining genetic material. We may use techniques that study all of your genes, only some of your genes and/or parts of your genetic material that do not have a currently known purpose or function. These techniques could include whole genome analysis in which all or most of your genetic code is studied and used to find the causes of disorders of brain development and related conditions. In some cases, we may use your blood to grow your cells in a dish. Blood cells grown this way can live indefinitely, giving a larger source of genetic material. Your samples will only be used to study the disorder in your family.

Confidentiality and Storage of Research Information, Samples and Data:

Information collected about you during this study will be given a unique code number and will not be put in your medical record. Your sample(s) and research data will be associated with your unique code only and stored without your name, medical record number or other identifying information. The information, samples and data will be accessible to the research study staff only and will be stored within the research laboratory in a secure and locked location and/or on a password-protected database at Boston Children's Hospital. Only the research study staff will be able to identify which sample(s), information and data belong to you and this link to your identity will not be shared with anyone outside of the study.

We may share your sample and/or research data with outside researchers who have an interest in the genetic cause(s) of the disorder of brain development in your family. In this case, your sample and/or research data would be released with the unique code only and not with your name or other identifying information. If at any time you would like to have your sample removed from storage in our laboratory, please let us know and it will be destroyed.

In order to allow researchers to share results, the National Institutes of Health (NIH) and other organizations have developed special data/information banks that collect and analyze DNA samples and results of whole genome studies. If provided to them, these central banks would store your genetic information and sample(s) and give them to other researchers to do more studies. Your sample(s) and data would be sent with your unique code number only; no identifiable information about you would ever be given to central banks. We do not think there will be further risks to your privacy and confidentiality by sharing your samples and whole genome information with these banks. However, we cannot predict how genetic information will be used in the future. There are many safeguards in place to protect your information and sample(s) while they are stored in these banks and used for research.

Risks and Discomforts:

Risks associated with a blood draw are minor discomfort and bruising. When possible, we will draw blood at the time of a clinically indicated procedure so that you will not need to have blood drawn only for research purposes.

Participants who may have an MRI as part of this study might feel claustrophobic while inside the MRI equipment and some might be bothered by the noise. Otherwise, MRI is generally a harmless imaging technique that does not involve using radiation, such as x-rays. MRI might not be possible for some individuals with metal implants, or for those who are not able to remain still during the MRI.

There is a chance that participation in this study could cause psychological distress. Some people involved in genetic studies have felt anxious about the chance of carrying an altered gene that places them at risk or that may be passed on to children. If you have 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.

You should also know that there might be social and economic disadvantages associated with the gathering of genetic information. You should know that our testing might find an inherited defective gene, which puts you at risk for a genetic disorder in the future. Genetic information given to the wrong source, could affect you and your family socially or economically (if an insurance company or employer acquired this genetic information). We will do our best to keep all information confidential and only with your permission would we make your information available to others. The results of research genetic tests will not be placed in your medical record. In this manner it will be unlikely that an insurance company or employer would ever learn of such results. You should know that we might detect instances of non-paternity (the discovery through genetic testing that the father is someone other than who it was thought to be), and such information may interfere with our analysis. This non-paternity information will be kept in the strictest confidence and will not be given to anyone.

In the event of an injury resulting directly from your participation in this research study, medical treatment will be provided if the injury is reported in a timely manner. Provision of such medical care does not imply any negligence or other wrongdoing on the part of the Hospital or any of the physicians or other personnel involved in the study. Where applicable, the Hospital reserves the right to seek payment from third-party payers for any medical care or services rendered. The Hospital has no program to provide you with any additional compensation as a result of any such injuries.

Potential Benefits:

Whereas we might find the genetic cause of the disorder of brain development in you/your child, it is possible we will not and you and your family might not directly benefit by participating in this study. We hope that information obtained from our research will help us better understand the genetic causes of disorders of brain development and eventually lead to new forms of treatment and diagnosis. For individuals having MRI as part of this study, the MRI could help provide the best possible diagnosis of your condition.

Research Result Possibilities and Reporting:

This research study is meant to find genes that are important for brain development and cannot study all possible diseases and genes. During the course of this research, we might find the genetic cause of the disorder of brain development in you/your child. Although we do not intend to, we might also find a genetic cause of a different disorder or uncover a risk of developing a disorder or disease in the future that is unrelated to the reason for your/your child's participation in this study.

You have the option of knowing if our study finds a genetic change in the sample collected from you/your child that, based on current scientific data and knowledge, could be (1) the cause of the disorder in you/your child or (2) the cause of another disorder or disease that could significantly affect your/your child's health or medical care. The latter would only include results that are proven to have a known significant effect on human health. You also have the option of not learning any results from this research.

Results from research genetic testing may take months or years to complete. It is possible that there will be no results from the research on your/your child's sample. If you wish to inquire into the progress of this research, you are welcome to do so at any time.

Since our research laboratory is not certified for reporting results to patients, we cannot give you results from our research genetic testing. However, if we find a result as described above, we may be able to have these results confirmed by a CLIA-certified clinical laboratory. A CLIA lab is allowed to release results from patient tests for clinical and diagnostic purposes. Result confirmation by a CLIA lab would involve the participation of your/your child's health care provider(s) and obtaining a new blood or saliva sample from you/your child. The result would be given to your health care provider and then to you with appropriate medical and genetic counseling. Your result could then be used for clinical and diagnostic purposes and could become part of your/your child's medical record. Testing in a CLIA lab could involve costs not covered by medical insurance.

Please read the three statements below and mark (□) next to the one that states your current wishes about results from this research study.

1. ☐ I **do not** want to learn about any results found about me/my child. Please **do not** contact me.

-or-

2. ☐ I want to learn only about results found about me/my child that could explain the condition that was the reason for my/my child's research participation (e.g. a brain malformation, epilepsy, mental retardation or autism).

-or-

3. ☐ I want to learn about results found about me/my child, including results that could (1) explain my/my child's condition (e.g. a brain malformation, epilepsy, mental retardation or autism) and/or (2) be the cause of another disorder or disease that could significantly affect my/my child's health or medical care but is unrelated to the reason for my/my child's research participation.

If you choose to be contacted, please indicate the name of the health care provider(s) that we should contact to discuss making arrangements with a CLIA lab. We will make every reasonable effort to get in touch with the person(s) you specify.

The health care provider(s) who can assist with confirmation of my/my child's research result:

Name: _____

Phone Number: _____

Fax Number: _____

Mailing Address: _____

Name: _____

Phone Number: _____

Fax Number: _____

Mailing Address: _____

You can change your mind about whether or not to receive results from this research at any time by contacting the Study Contacts listed below. The Study Contacts are also available to discuss your options further at your request.

Alternatives:

Participation in this research study is completely voluntary. You should not feel any pressure to participate. If you do not want to participate or you withdraw from the study, it will not interfere with any future care you or your family receives at this institution.

The National Institutes of Health has issued a Certificate of Confidentiality for this research. This adds special protection for the research information and specimens that may identify you. The researchers may not disclose information that may identify you, even under a court order or subpoena unless you give permission. However, a Certificate of Confidentiality does not prevent researchers from disclosing information about you if required by law (such as to report child abuse, communicable diseases or harm to self or others); if you have consented to the disclosure (such as for your medical treatment); or if it is used for other research as allowed by law. In addition the Certificate cannot be used to refuse a request if a governmental agency sponsoring the project wants to audit the research. Any research information that is placed in your medical record would not be covered under this Certificate. The Certificate will not be used to prevent disclosure for any purpose you have consented to in this informed consent document. The Certificate does not stop you from voluntarily releasing information about yourself or your involvement in this research. If others obtain your written consent to receive research information, then the researchers may not use the Certificate to withhold that information.

What do I need to know about Privacy and Confidentiality?

You/your child's health information is protected by a law called the Health Information Portability and Accountability act (HIPAA). In general, anyone who is involved in this research including those funding and regulating the study may see the data, including information about you. For example, the following people might see information about you:

- Research staff at Boston Children's Hospital involved in this study.
- Medical staff at Boston Children's Hospital directly involved in your care that is related to the research or arises from it.
- Other researchers and centers that are a part of this study, including people who oversee research at that hospital.
- People at Boston Children's Hospital who oversee, advise, and evaluate research and care. This includes the ethics board and quality improvement program.
- People from agencies and organizations that provide accreditation and oversight of research.
- People that oversee the study information such as data safety monitoring boards, clinical research organizations, data coordinating centers, and others.
- Sponsors or others who fund the research, including the government or private sponsors.
- Companies that manufacture drugs or devices used in this research.
- Federal and state agencies that oversee or review research information, such as the Food and Drug Administration, the Department of Health and Human Services, the National Institutes of Health, and public health and safety authorities.

- People or groups that are hired to provide services related to this research or research at Boston Children's Hospital, including services providers, such as laboratories, and others.
- Your health insurer for portions of the research and related care that are considered billable.

If some law or court requires us to share the information, we would have to follow that law or final ruling.

Some people or groups who get your health information might not have to follow the same privacy rules. Once your information is shared outside of Boston Children's Hospital, we cannot promise that it will remain private. If you/your child decide to share private information with anyone not involved in the study, the federal law designed to protect privacy may no longer apply to this information. Other laws may or may not protect sharing of private health information. If you have a question about this you may contact the Boston Children's Hospital Privacy Office at 857-218-4680 which is set up to help you understand privacy and confidentiality.

Because research is ongoing we cannot give you an exact time when we will destroy this information. Researchers continue to use data for many years so it is not possible to know when they will be done.

We will also create a code for the research information we collect about you so identifying information will not remain with the data and will be kept separately. The results of this research may be published in a medical book or journal or be used for teaching purposes. However your name or identifying information will not be used without your specific permission.

Your privacy rights

If you or your child do not want to participate in this study, you do not have to. If you do want to participate, however, you must sign this form.

If you do not sign this form, it will not affect your care or your child's care at Boston Children's Hospital now or in the future and there will be no penalty or loss of benefits. You/your child can withdraw from the study and end your permission for Boston Children's Hospital to use or share the protected information that was collected as part of the research; however you cannot get back information that was already shared with others. Once you remove your permission, no more private health information will be collected. If you wish to withdraw your health information will need to do so in writing.

You/your child may have the right to get some the information that was shared with others for research, treatment or payment. This information is available after the study analysis is done. To request the information, please contact the Hospital's Privacy Officer at 857-218-4680.

Contact Information

I understand that I may use the following contact information to reach the appropriate person/office to address any questions or concerns I may have about this study. I know:

I can call…	At		If I have questions or concerns about
Investigator: **Christopher Walsh, MD**	Phone:	**617-919-2923**	▪ General questions about the study ▪ Research-related injuries or emergencies ▪ Any research-related concerns or complaints
Study Contacts: **Jennifer Partlow, MS** **Abbe Lai, MS**	Phone: Phone:	**617-919-2865** **617-919-4371**	▪ General questions about the study ▪ Research-related injuries or emergencies ▪ Any research-related concerns or complaints
Office of Clinical Investigations	Phone:	**617-355-7052**	▪ Rights of a research subject ▪ Use of protected health information. ▪ Compensation in event of research-related injury ▪ Any research-related concerns or complaints. ▪ If investigator/study contact cannot be reached. ▪ If I want to speak with someone other than the Investigator, Study Contact or research staff.

Documentation of Informed Consent and Authorization

- I have read this consent form and was given enough time to consider the decision to participate in this study.
- This research study has been satisfactorily explained to me, including possible risks and benefits.
- All my questions were satisfactorily answered.
- I understand that participation in this research study is voluntary and that I can withdraw at any time.
- I am signing this consent form prior to participation in <u>any</u> research activities.
- I give permission for my/my child's participation in this research study and for the use of associated protected health information as described above (HIPAA).

<u>Parent/Legal Guardian Permission (if applicable)</u>

If the child to be involved in this research study is a <u>foster child</u> or a <u>ward of the state</u> please notify the researcher or their staff who is obtaining your consent.

■ _____ _____ _____

Date (MM/DD/YEAR) Signature of **Parent** or **Legal Guardian** Relationship to child

<u>Child Assent (if applicable)</u>

■ _____ _____

Date (MM/DD/YEAR) Signature of **Child/Adolescent Subject**

■ If child/adolescent's assent is **<u>not</u>** obtained above, please indicate reason below (check one):

☐ Assent is documented on a <u>separate</u> IRB-approved assent form
☐ Child is too young
☐ Other reason (e.g. sedated), please specify: _____

Adult Subject (if applicable)

■ _____ _____
Date (MM/DD/YEAR) Signature of **Adult Subject** *(18+ years)*

Investigator or Associate's Statement & Signature

- I have fully explained the research study described above, including the possible risks and benefits, to all involved parties (subject/parents/legal guardian as applicable).
- I have answered and will answer all questions to the best of my ability.
- I will inform all involved parties of any changes (if applicable) to the research procedures or the risks and benefits during or after the course of the study.
- I have provided a copy of the consent form sign by the subject/ parent / guardian and a copy of the hospital's privacy notification (if requested)

■ _____ _____
Date (MM/DD/YEAR) Signature of **Investigator or Associate**

Witness Statement & Signature

A witness must be present for the entire consent process in the following situations (please check the appropriate box)

☐ The individual cannot read and this consent document was read to the subject or legal representative, **or**
☐ The individual has certain communication impairments that limit the subject's ability to clearly express consent **or**
☐ Situations where the IRB requests a witness be present: please specify _____

I confirm that the information in this consent form was accurately explained to the subject, parent or legally authorized representative, the individual appeared to understand the information and had the opportunity to ask questions, and that informed consent was given freely.

☐ _____ _____
Date (MM/DD/YEAR) Signature of **Witness**

Or

☐ The individual is not English speaking and, through an interpreter, a short form consent document was presented orally to the subject or legal representative and this consent document serves as the summary for such consent.

I confirm that the information in this consent form was presented orally to the subject, parent or legally authorized representative, in a language they could understand and the individual had the opportunity to ask questions.

☐ _____ _____
Date (MM/DD/YEAR) Signature of **Witness and Interpreter**