Boston Children's Hospital

RESEARCH CONSENT FORM

Use Plate or Print:

Subject Name:

DOB:

Gender:

Protocol Title: Genetics of Epilepsy and Cognitive Disorders

Principal Investigator: Christopher Walsh

This consent form gives you important information about a research study. A research study helps scientists and doctors learn new information to improve medical practice and patient care. Please read this consent form carefully and take your time making a decision. Participation in this research is voluntary. You may choose not to take part in this research study or may choose to leave the research study at any time. Your decision will not impact the clinical care you receive at Boston Children's Hospital. This consent form gives you detailed information about the procedures, risks and benefits of the study, and may contain words that you do not understand. Please ask questions about anything you do not understand. We encourage you to talk to others (for example, your friends, family, or other doctors) before you decide to participate in this research study.

Please check one of the following:

_____ You are an adult participant in this study.

_____ You are the parent or guardian granting permission for a child in this study.

If the participant is a child/dependent, the use of "you/your/me/my" in this form refers to that individual.

Study Summary

- This study aims to identify and understand genes important for brain development and function.
- You are invited to participate in this study since you or a family member has a disorder of brain development and/or function. Participation is voluntary and will not impact any care you or your family receives at this institution.
- Participation involves sharing your health, family history and genetic information, as well as biological sample(s). With your samples, we might study some or all of your genes and make cell lines to investigate how your genes and cells are working.
- Some risks of this study include possible discomfort and bruising from a blood draw, psychological distress about genetic information, and loss of confidentiality.
- Benefits could include finding the genetic cause of the brain disorder in you/your family and/or advancing scientific knowledge or medical practice in the future but there might be no benefit. Genetic results about you might be available from this study. You can decide if you wish to receive certain results, which must be confirmed through clinical testing.
- We might share your anonymous sample and/or genetic and clinical data with other scientists and researchers to study your or other health conditions. Your privacy is important and this form will explain how we manage your personal information and samples.

Why is this research being done and how are individuals selected for the study?

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 intellectual disability and autism, and other related conditions. Many genetic causes of these conditions remain unknown. This research aims to discover the genes underlying these disorders and, in some cases, study known genetic causes to further characterize the genes important to brain development and function.

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. 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 and understand the many genes important for brain development, and we continue to enroll individuals on an ongoing basis.

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.

Who is conducting this research study, and where is it being conducted?

Christopher Walsh, MD, PhD is the lead investigator of this study and the research is done at Boston Children's Hospital. Dr. Walsh also collaborates with other researchers and clinicians around the world to help achieve the goals of the study.

What do I have to do if I am in this research study?

If you choose to participate, you may 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 for medical information about you. We may continue to obtain updates on your health from your medical record for as long as you participate in this study. 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, DNA, RNA) and the products your genes make (molecules and proteins). 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 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, or a hair sample from pulling a few hairs from your head. If a previously or subsequently collected DNA, blood, tissue or other biological sample or genomic sequence data exists, that is not needed for clinical purposes, we may request this as part of this research.

How will my samples and information be stored, used and shared?

Identifiable sample(s) and identifiable private information will be collected from you during this study, and will be given a unique research code. 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.

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 examine the causes of disorders of brain development and related conditions.

Cell lines and induced pluripotent stem cells: The blood or tissue collected from you in this research may be used to create a "cell line" that can be grown in the laboratory. A cell line can continue to grow and make more cells indefinitely. This allows researchers to have an unlimited supply of your cells in the future without asking for more samples from you. We may also use the cells taken from your blood to create a type of cell known as a "pluripotent stem cell". Stem cells can be used to create other types of cells and tissue, including nerve and brain cells. Your cells might be used to study genetic changes and the processes in the cells that could be affected by those changes. The researchers will use your cells to try to learn more about disorders of brain development.

Future use and sharing of samples and information: The samples and/or information collected from you during this study may be shared with collaborating researchers who also have an interest in the disorder of brain development in your family. They may also be shared for future research on many types of diseases or conditions. If we distribute your samples and/or information to other researchers or institutions, your samples and/or information will be labeled with a research code without identifiers so that you cannot be identified. No additional consent will be requested for the future use of your samples or information.

If you have questions about storing samples or would like to request that samples be removed from storage, please let us know. It is not always possible to remove samples from storage or to retrieve samples from which identifiers have been removed and/or that have already been sent to other investigators.

Additionally, as this research is funded by the National Institutes of Health (NIH), it is subject to the federal Genomic Data Sharing Policy. This policy requires genetic studies to obtain participants' consent for their sample and genetic and phenotypic (clinical) data to be used for future research and shared broadly. In order to allow researchers to share results, the NIH and other central repositories have developed special sample/data (information) "banks" that collect the results and analyze samples/data from research studies, including genetic studies. These central banks may also analyze and store samples and health information from research conducted by Boston Children's Hospital. These central banks will store your genetic and health information and/or samples and give them to other qualified and approved researchers to do more studies on many types of diseases or conditions. We do not think that there will be further risks to your privacy and confidentiality by sharing your health information, samples and/or genetic information with these banks. However, we cannot predict how genetic information will be used in the future. The samples and data will be sent with only your research code number attached. Your name or other directly identifiable information will not be given to these central banks. There are many safeguards in place to protect your privacy.

What are the risks of this research study? What could go wrong?

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.

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.

Our testing might find a gene which may put you or a relative at risk for a genetic disorder in the future. There might be social and economic disadvantages associated with genetic information. For example, genetic information provided to the wrong source could affect you and your family. A U.S. law called the Genetic Information Nondiscrimination Act (GINA) generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. Under this law, health insurance companies, group health plans, and most employers may not request your genetic information that we get from this research. For more information about GINA, please see: http://www.eeoc.gov/laws/types/genetic.cfm. This law does not protect information for being used when applying for life or long term care insurance. We will do our best to keep all information confidential and only with your permission would we share this information with others.

You should be aware that because we are testing family members, we may find out that someone else might have fathered a child, or that a child had been adopted. If you wish, you may let us know in confidence if this is a possibility. In all cases, this information will be kept in the strictest confidence and will not be shared with you or anyone outside of the research staff.

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.

What are the benefits of this research?

We might find the genetic cause of the disorder of brain development in you or your family member, if it is not already known. However, it is also possible we will not find the genetic cause 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.

Research Result Possibilities and Reporting

This research is meant to identify and study genes that are important for brain development but cannot study all possible diseases and genes. During the course of this research, we might find the genetic cause of your disorder of brain development, if it is not already known. 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 participation in this study.

You have the option of knowing if our study finds a genetic change in the sample collected from you that, based on current scientific data and knowledge, could be (1) the cause of your disorder, if not already known, or (2) the cause of another disorder or disease that could significantly affect your 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. Results other than genetic testing results are not anticipated from this study and it is possible that there will be no results from the research on your 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 health care provider(s) and obtaining a new blood or saliva sample. 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 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 regardless of the reason for my research participation. Please **do not** contact me with results.

-or-

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

-or-

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

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.

Other information that may help you

Boston Children's Hospital is interested in hearing your comments, answering your questions, and responding to any concerns regarding clinical research. If you have questions or concerns, you may email IRB@childrens.harvard.edu or call (617) 355-7052 between the hours of 8:30 and 5:00, Monday through Friday.

Who may see, use or share your health information?

Your participation in this research study may be noted in your electronic medical record. If you do not have an existing medical record, one may be created. Information placed in your medical record may include the informed consent or the results of tests or assessments we collect during the research. This could be viewed by staff from Boston Children's Hospital, study monitors and others who provide oversight of the study. Everyone who sees this information is required to keep it confidential in accordance with hospital policies and laws. Information about your research participation may not be given to anyone unaffiliated with Boston Children's Hospital in a way you can be identified unless we obtain your permission, or it is permitted or required by law.

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 should you know about HIPAA and confidentiality?

Your 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;
- And/or 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 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 Officer 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 want to participate in this research study, you must sign this form. If you do not sign this form, it will not affect your care at Boston Children's Hospital now or in the future and there will be no penalty or loss of benefits. You 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, please contact the research team. You may have the right to find out if information collected for this study was shared with others for research, treatment or payment. You may not be allowed to review the information, including information recorded in your medical record, until after the study is completed. When the study is over, you will have the right to access the information again. 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 Neil, MS** **Walsh Lab Research Staff**	Phone: **617-919-2865** **617-919-4795**	▪ 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 any 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).

Parent/Legal Guardian Permission (if applicable) If the child to be involved in this research study is a **foster child** or a **ward of the state** 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

Child Assent (if applicable)

■ _____ _____
Date (MM/DD/YEAR) Signature of **Child/Adolescent Participant**

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

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

Adult Participant (if applicable)

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

Adult Participant: If decisionally impaired (if applicable)

Legal Authorized Representative/Guardian: I give permission for the person I am authorized to represent to participate in this research and for the use of associated protected health information as described above (HIPAA).

■ _____ _____ _____
Date (MM/DD/YEAR) Signature of **Legal Guardian** Print Name

■ Relationship to Participant * *(This order must be followed. If there is a court appointed guardian, this is who needs to provide consent. If not, a health care proxy, followed by durable power of attorney and lastly, family members)*

☐ Court-Appointed Guardian
☐ Health Care Proxy (Attach Proxy, ensure there is express authority to make health care decisions including research.)
☐ Durable Power of Attorney (POA) (Durable POA may be limited to specific areas. Attach Durable POA, ensure that it covers research.)
☐ Family Member/Next of Kin (order of preference: spouses, parents, adult children) Relationship_____

Adult Assent (if applicable)

■ _____ _____
Date (MM/DD/YEAR) Signature of **Adult Participant**

☐ CHECK if Adult Participant's assent **not** obtained above, and specify reason below:

_____

Research Investigator or Associate's Statement & Signature

- I have fully explained the research described above, including the possible risks and benefits, to all involved parties (participant/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 research.
- I have provided a copy of the consent form sign by the participant/parent/guardian and a copy of the hospital's privacy notification (if requested)

■ _____ _____

Date (MM/DD/YEAR) Signature of **Research 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 participant or legal representative, **or**

☐ The individual has certain communication impairments that limit the participant'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 participant, 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 participant 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 participant, 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**