

**UNIVERSITY OF CALIFORNIA, SAN FRANCISCO
 CONSENT TO PARTICIPATE IN A RESEARCH STUDY**

Epileptic Encephalopathies: Clinical and Genetic Predictors of Outcomes and Therapeutic Insights

This is a research study. The study doctor Elliott H. Sherr, M.D., Ph.D. from the Department of Neurology and/or a research coordinator will explain this study to you.

Research studies include only people who choose to take part. Please take your time to make your decision about your child’s participation in this study, and discuss your decision with your family or friends if you wish. If you have any questions, you may ask the researchers.

Your child is being asked to take part in this study because:

1. He or she has been given a clinical diagnosis of an epileptic encephalopathy such as Infantile Spasms (IS) or Lennox-Gastaut Syndrome (LGS).

Why is this study being done?

Dr. Sherr and his colleagues at UCSF conduct clinical research seeking to advance the understanding of epileptic encephalopathies. With these investigations, we are trying to understand the genetic predictors of such disorders and their relationship with long-term clinical outcomes.

How many people will take part in this study?

About 150 people with IS or another epileptic disorder will take part. Both of their biological parents and siblings will be invited to enroll and participate. Overall, about 900 patients and controls will be enroll in the study.

What will happen if my child takes part in this research study?

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

1. The researchers will gather information about your child from your child’s medical record.
2. You will bring your child to UCSF’s medical center or Pediatric Clinical Research Center (PCRC) and your child will give a blood sample. The blood will be drawn by putting a needle into a vein in your child’s arm. One to two small tubes of blood (approximately one tablespoon) will be taken. This will take about five minutes. If your child is not able to visit UCSF to donate the blood sample, we will provide you with a kit that you can take to your local clinical lab or doctor’s office. The kit will contain tubes, instructions, and shipping supplies, and a prepaid return shipping label.

3. We are also interested in studying tissue obtained during medical or research procedures, for example a skin or muscle biopsy. If your child will be receiving a biopsy as part of their care, we would like to obtain any tissue that would otherwise be discarded from the medical or research procedure. Additionally, if your child wishes to receive a biopsy for research purposes, a 3 mm skin sample will be obtained using a standard punch biopsy needle. Analysis of the tissue can often teach us important information about diseases that cannot be learned from studying blood and/or saliva alone. Your child may also wish to donate a hair sample for research. A separate skin biopsy and hair donation consent form will be signed if your child agrees to this part of the study.
4. Your child will be asked to complete an evaluation with a psychologist at your home where the psychologist will travel. This entails a cognitive and behavioral evaluation with an age-appropriate subset of instruments to measure verbal, nonverbal, social-communication and motor development skills. During the evaluation, the psychologist will ask your child questions to assess your child's thinking at the time of the test. You may also be asked to provide additional information about your child's health and behavior by completing questionnaires and/or a telephone interview. The total estimated testing time is 2-8 hours.
5. We are able to release a limited amount of data that we have collected to participants. We can provide the scaled scores from the standardized neuropsychological tests performed during the evaluation. In neither case should this substitute for standard medical care. However, if we identify important and treatable clinical information during our study, we will inform you in a timely manner and help you quickly identify medical assistance.

How long will my child be in the study?

The research activities outlined above can be completed in 1- 2 visits. The total time required is not expected to exceed 8 hours.

Can my child stop being in the study?

If you first agree to let your child participate and then change your mind, you are free to withdraw your child from the study and discontinue your child's participation at any time. If you choose to tell the researchers why you are leaving the study, your reasons for leaving may be kept as part of the study record. Your decision will not affect your child's ability to receive medical care and your child will not lose any benefits to which your child would otherwise be entitled. Also, the investigators or other member of the research team may stop your child from taking part in this study at any time if he or she believes it is in your child's best interest, if your child or you do not follow the study rules, or if the study is stopped.

What risks can my child expect from being in the study?

Drawing blood may cause temporary discomfort from the needle stick. Other risks associated with having blood drawn are slight but may include fainting or feeling light-headed, bruising, and infection. There is also a small risk that your child might feel distressed or nervous about having blood drawn (venipuncture).

You might feel worried about the possibility of your child's personal information and/or participation in this study not being kept confidential. The risk of unwanted sharing of health information will be minimized by ensuring that all study records that identify your child will be kept completely confidential. A unique study subject number will be used on sample tubes as well as databases containing your child's follow-up testing and questionnaire information. Only select members of the primary research team will have access to the original data. The information you and your child provide will be stored in a locked file cabinet.

There may be risks that are unforeseen and no one knows about yet. The researchers will let you know if they learn anything that might make you change your mind about your child's participation in the study.

For more information about risks, ask one of the investigators or research coordinators.

Are there benefits to taking part in the study?

There will be no direct benefit to you from participating in this study. However, the information that you and your child provide may help health professionals better understand epilepsy with the goal of linking genetics and baseline clinical variables to long-term clinical outcomes, which may support the development of novel therapeutic strategies.

What other choices do I have if my child does not take part in this study?

You are free to choose not to have your child participate in the study. If you and/or your child decide not to take part in this study, there will be no penalty to you or your child. Your child will not lose any of his or her regular benefits, and your child can still get care from our institution the way he or she usually does.

Will my child's medical information be kept private?

We will do our best to protect the information we collect about your child. Information which identifies your child will be kept secure and restricted. However, your child's personal information may be given out if required by law. If information from this study is published or presented at scientific meetings, your child's name and other identifiers will not be used. Information which identifies your child will be destroyed when this research is complete. Your child's personal information may be given out if required by law. The following information about you in your medical and research records:

- UCSF's Committee on Human Research

- Government agencies involved in keeping research safe for people, such as the Food and Drug Administration (FDA).

Sensitive research information: Participation in research may involve a loss of privacy, but information about your child will be handled as confidentially as possible. There is a risk that information about taking part in a study about ASD may influence insurance companies and/or employers regarding your child’s health. However, no information obtained in this study will be placed in your child’s medical records nor will it be provided to any insurance entity or place of employment, except in the situation described as follows: your child’s participation in this study may be noted in your child’s medical record at UCSF. If your child does not already have a medical record at UCSF, one will be created for them, since they will be completing some or all of the research activities associated with this study at UCSF’s medical center and Pediatric Clinical Research Center (PCRC). Your child’s consent forms, documentation of the blood draw procedure, and/or results of the neuropsychological assessments may be included in this record. Other doctors at UCSF may become aware of your child’s participation. However, hospital regulations require that all health care providers treat information in medical records confidentially.

What are the costs of taking part in this study?

There will be no cost to you for any of the study procedures. In the event that your child has his/her blood drawn outside of UCSF for the purposes of this research study, we will reimburse you up to \$30.00 to cover the costs of the blood draw at an offsite facility. We will also reimburse you for parking costs if your child and you visit UCSF to complete any of the research activities described under the section, “What will happen if my child takes part in this research study?”

Will my child be paid for taking part in this study?

In addition to the study reimbursements outlined above, your child will not be paid for participating in this study.

What happens if my child is injured because of taking part in this study?

It is important that you tell Dr. Sherr if you feel that your child has been injured because of taking part in this study. You can tell the doctor in person or call (415) 502-8039.

Treatment and Compensation for Injury: If your child is injured as a result of being in this study, treatment will be available. The costs of the treatment may be covered by the University of California, depending on a number of factors. The University does not normally provide any other form of compensation for injury. For further information about this, you may call the office of the Committee on Human Research at 415- 476-1814.

What are my child’s rights if they take part in this study?

Having your child take part in this study is your choice. You may choose to have your child take part or not to take part in the study. If you decide to let your child take part in this study, you are free to leave the study at any time. No matter what decision you make, there will be no penalty to you or your child and your child will not lose any of their regular benefits. Leaving the study will not affect your child’s medical care. Your child can still get their medical care from our institution. We will tell you about new information or changes in the study that may affect your willingness to have your child continue in the study. In the case of injury resulting from this study, you will not lose any of your or your child’s legal rights to seek payment by signing this form.

Who can answer my questions about the study?

You can talk to the researchers about any questions, concerns, or complaints you have about this study by calling the research staff or Dr. Sherr at (415) 502-8039.

If you wish to ask questions about the study or your child’s rights as a research participant to someone other than the researchers or if you wish to voice any problems or concerns you may have about the study, please call the Office of the Committee on Human Research at 415-476-1814.

Multi-site Collaboration

For this project we are collaborating with Columbia Univeristy (Drs. Melodie Winawer and Dale Hesdorffer), Albert Einstein College of Medicine (Dr. Shlomo Shinnar), Emory University (Dr. Michael Epstein), and Harvard Univeristy (Dr. Sarah Spence). We will ask for your consent to have your/your child’s personal health information shared between this study and the previously mentioned research programs at the end of this form.

OPTIONAL:Use of Tissue for Research

About Using Tissue for Research

As part of your child’s participation in this research, a tissue sample (blood) will be collected. We would like to keep some of the tissue that is leftover for future research. If you agree, the tissue specimen(s) will be kept in storage in what is called a “tissue bank.” Dr. Sherr’s tissue bank at UCSF will house tissue specimens collected for the purposes of this research study.

We do not know for sure if your child’s tissue specimen(s) or information will be used, but they might be used in research about neurological, psychiatric and behavioral disorders. Your child’s specimen(s) and any information about your child will be kept until it is used up or destroyed. If you decide later that you do not want your child’s tissue specimen(s) and information to be used

for future research, you can notify the investigator and/or a member of the research team by calling (415) 502-8039 and/or in writing, and any remaining identifiable tissue specimen(s) and information will be destroyed if they are no longer needed for your child's care. All written requests should be directed to Elliott Sherr, M.D., Ph.D. at 675 Nelson Rising Lane, Box 3206, San Francisco, CA 94158. However, if any research has already been done using portions of your child's tissue specimen(s), the data will be kept and analyzed as part of those research studies. Reports about research done with your child's tissue will not be given to you or your child's doctor. These reports will not be put in your child's health record. The research will not have an effect on your child's care.

Things to Think About

- The choice to let us keep the leftover tissue specimen(s) for future research is up to you. No matter what you decide to do, it will not affect your child's care.
- If you decide now that your child's tissue specimen(s) can be kept for research, you can change your mind at any time. Just contact us and let us know that you do not want us to use your child's tissue. Then any tissue that remains will no longer be used for research.
- Sometimes tissue specimens are used for genetic research (about diseases that are passed on in families) including whole exome and/or whole genome sequencing. Even if we use your child's tissue for genetic research, we will not put the results in your child's medical record.
- We may give your child's tissue specimen(s) and certain medical information about your child (for example, diagnosis, age, developmental history, etc.) to other scientists or companies not at UCSF, including a government health research database, but we will not give them your child's name, address, phone number, or any other information that would identify your child. Reports about any research will not be given to you or your child's doctor.
- You will not be charged for donating your child's tissue specimen(s). Your samples or data may be used to develop new products, tests or discoveries. In some instances, these may have potential commercial value. You will not receive any payment or financial benefit from any products, tests or discoveries.
- Your child's personal health information cannot and will not be used for additional research without additional approval from either you or a review committee.

CERTIFICATE OF CONFIDENTIALITY

To help us protect your privacy, we have obtained a Certificate of Confidentiality from the National Institutes of Health. With this Certificate, the researchers cannot be forced to disclose information that may identify you, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings.

Exceptions: A Certificate of Confidentiality does not prevent researchers from voluntarily disclosing information about you, without your consent. For example, we will voluntarily disclose information about incidents such as child abuse, and intent to hurt yourself or others. In addition, a Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. If an insurer, employer, or other person obtains your written consent to receive research information, then the researchers may not use the Certificate to withhold that information. Finally, the Certificate may not be used to withhold information from the Federal government needed for auditing or evaluating Federally funded projects or information needed by the FDA.

Benefits

There is no direct benefit to you or your child for donating your child's tissue specimen(s) and agreeing to have your child's tissue specimen(s) stored and potentially used for research. However, the benefits of research using tissue include learning more about what causes neurological, psychiatric and behavioral disorders and other diseases, how to prevent them, and how to treat them.

The greatest risk to your child is the release of information from your child's health records. We will do our best to make sure that your child's personal information will be kept private. The chance that this information will be given to someone else is very small.

Making Your Choice

Please read each sentence below and think about your choice. **After reading each sentence, please initial either the "Yes" or "No" box.** If you have any questions, please talk to your doctor or nurse, or call our research review board at 415-476-1814. No matter what you decide to do, it will not affect your child's care.

YES	NO	1. My child's tissue may be kept for use in research to learn about, prevent, or treat neurological, psychiatric, behavioral disorders and/or other health problems
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YES	NO	2. My child's tissue may be kept for use in research to learn about, prevent or treat other health problems (for example: diabetes, Alzheimer's disease, or heart disease).
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YES	NO	3. My child's tissue may be kept for use in genetic research, including whole exome and/or whole genome sequencing.
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YES	NO
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4. Someone may contact me in the future to ask me about whether or not I'm interested in having my child take part in more research.

YES	NO
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5. I allow the sharing of my child's personal health information between Dr. Sherr and his collaborators as outlined in the "Multisite Collaboration" section of this consent form.

CONSENT

You have been given copies of this consent form and the Experimental Subject's Bill of Rights to keep. You will be asked to sign a separate form authorizing access, use, creation, or disclosure of health information about your child.

PARTICIPATION IN RESEARCH IS VOLUNTARY. You have the right to decline to participate or to withdraw at any point in this study without penalty or loss of benefits to which your child is otherwise entitled.

1. Parent/Legal Guardian Signature

The person being considered for this study is unable to consent for him or herself because he or she is a minor. By signing below, you are giving your permission for your child to be included in this study.

_____	_____	_____
Date	Signature of Parent/Legal Guardian	Print Name
	_____	_____
	Relationship to Minor Participant	Name of Minor Participant

2. Study Staff Signature

_____	_____	_____
Date	Signature of Person Obtaining Consent	Print Name of Person Obtaining Consent

