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 participating 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.

You are being asked to take part in this study because:

1. Your child 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 infantile spasms (IS) as well as other epileptic disorders. 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 I take part in this research study?

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

1. The researchers will gather information about you from a brief telephone interview or online questionnaire.
2. You will visit UCSF’s medical center to give a blood sample. The blood will be drawn by putting a needle into a vein in your arm. Two tubes of blood (approximately one tablespoon) will be taken. This will take about five minutes. If you are 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. You will be asked to complete questionnaires and/or a telephone interview regarding your social, verbal, and behavioral traits. This is expected to take 1-2 hours.
4. We are able to release a limited amount of data that we have collected to participants. 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 I be in the study?

The research activities outlined above will require up to three hours of your time.

Can I stop being in the study?

If you first agree to participate and then change your mind, you are free to withdraw from the study and discontinue your 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 ability to receive medical care and you will not lose any benefits to which you would otherwise be entitled. Also, the investigators or other member of the research team may stop you from taking part in this study at any time if he or she believes it is in your best interest, if you do not follow the study rules, or if the study is stopped.

What risks can I 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 you might feel distressed or nervous about having blood drawn (venipuncture).

You might feel worried about the possibility of your 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 you will be kept completely confidential. A unique study subject number will be used on sample tubes as well as databases containing your follow-up testing and questionnaire information. Only select members of the primary research team will have access to the original data. The information you 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 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 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 I do not take part in this study?

You are free to choose not to participate in the study. If you decide not to take part in this study, there will be no penalty to you. You will not lose any of your regular benefits, and you can still get care from our institution the way you usually do.

Will my information be kept private?

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

- 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 you 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 health. However, no information obtained in this study will be placed in your medical records nor will it be provided to any insurance entity or place of employment, except in the situation described as follows: your participation in this study may be noted in your medical record at UCSF. If you do not already have a medical record at UCSF, one will be created for you if you complete some or all of the research activities associated with this study at UCSF’s medical center. Your consent forms and documentation of the blood draw procedure may be included in this record. Therefore, other doctors at UCSF may become aware of your 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 you have your 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. In addition to reimbursing up to $30.00 for costs of the blood draw outside UCSF, we will also reimburse you for the cost of parking during your visit(s) to UCSF for the cognitive and behavioral assessments.

Will I be paid for taking part in this study?

In addition to the study reimbursements outlined above, you will not be paid for participating in this study.
What happens if I am injured because of taking part in this study?

It is important that you tell Dr. Sherr if you feel that you have 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 you are 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 rights if I take part in this study?

Taking part in this study is your choice. You may choose to take part or not to take part in the study. If you decide to 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 and you will not lose any of your regular benefits. Leaving the study will not affect your medical care. You can still get medical care from our institution. We will tell you about new information or changes in the study that may affect your willingness to continue in the study. In the case of injury resulting from this study, you will not lose any of your 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 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 University (Drs. Melodie Winawer and Dale Hesdorffer), Albert Einstein College of Medicine (Dr. Shlomo Shinnar), Emory University (Dr. Michael Epstein), and Harvard University (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 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 tissue specimen(s) or information will be used, but they might be used in research about neurological, psychiatric and behavioral disorders. Your specimen(s) and any information about you will be kept until it is used up or destroyed. If you decide later that you do not want your 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 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 tissue specimen(s), the data will be kept and analyzed as part of those research studies. Reports about research done with your tissue will not be given to you or your doctor. These reports will not be put in your health record. The research will not have an effect on your 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 care.
- If you decide now that your 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 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 tissue for genetic research, we will not put the results in your medical record.
- We may give your tissue specimen(s) and certain medical information about you (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 name, address, phone number, or any other information that would identify you. Reports about any research will not be given to you or your doctor.
- You will not be charged for donating your 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 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 for donating your tissue specimen(s) and agreeing to have your 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 you is the release of information from your health records. We will do our best to make sure that your 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.

<table>
<thead>
<tr>
<th>YES</th>
<th>NO</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. My tissue may be kept for use in research to learn about, prevent, or treat neurological, psychiatric, behavioral disorders and/or other health problems</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>YES</th>
<th>NO</th>
</tr>
</thead>
<tbody>
<tr>
<td>2. My 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).</td>
<td></td>
</tr>
</tbody>
</table>
3. My tissue may be kept for use in genetic research, including whole exome and/or whole genome sequencing.

4. Someone may contact me in the future to ask me about whether or not I’m interested in taking part in more research.

5. I allow the sharing of my 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 you.

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 you are otherwise entitled.

1. Participant Signature

I agree to participate by signing below.

Date ___________________________ Participant’s Signature ___________________________ Print Name ___________________________

2. Study Staff Signature

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