

**UNIVERSITY OF CALIFORNIA, SAN FRANCISCO
CONSENT TO DONATE SPECIMENS FOR FUTURE RESEARCH**

Study Title: *Disorders of Cerebral Development: A Phenotypic and Genetic Analysis*

This is a request that you or your child donate specimens for medical research. The principal investigator, Dr. Elliott H. Sherr from the Department of Neurology and/or one of his research coordinators will explain this research to you.

Medical research includes only people who choose to take part. Take your time to make your decision about participating. You may discuss your decision with your family and friends and with your health care team. If you have any questions, you may ask the researchers.

You or your child are participating in this study because you/your child or a close relative has a disorder of brain development that is visible on an MRI (or other imaging study) and/or a disorder of brain development that has been clinically diagnosed, such as autism, epilepsy or cerebral palsy. We are collecting blood samples from participants as well as certain family members as a part of the genetic analysis. In addition, we are collecting other tissue samples including those obtained during a medical or research procedure(s) performed by your personal physician or the principal investigator Dr. Elliott Sherr.

Why is this research being done?

The purpose of this study is to learn about brain development and disorders that occur in this process. They would like to understand problems and issues associated with having these conditions and they would like to investigate the potential genetic and environmental causes. We understand that medical procedures are stressful and taking the extra step to make a tissue donation may be a difficult decision. We will work with you/your child and your/your child's treating physician to make this process as simple as possible. Your/your child's tissue will be saved in what is called a "tissue bank" for possible future research.

Another goal of this study is to convert skin cells from patients with neurodevelopmental disorders into other cell types that can be studied in the laboratory. If you participate in this part of the study, a separate consent form will cover the procedures involved in this. To achieve this we will convert cells from the skin into induced pluripotent progenitor (iPS) cells. A pluripotent cell can create all other kinds of cell types. iPS cells are a type of pluripotent stem cell that is artificially derived by introducing specific genes into cells. We will grow these pluripotent stem cells into other types of cells, such as neurons, similar to those that make up other tissues in the body. We hope that these studies will help us identify what is different in the cells of patients in this study.

- This project is funded by the National Institutes of Health, March of Dimes, Weston Havens, and Pfizer.

How many people will take part in this research?

About 900-1000 individuals with disorders of cerebral development, their parents and other significant relatives will take part in this research study. We are unable to predict the number of people who will be donating tissue for this research.

What will happen if I agree to donate my specimens?

If you agree to let researchers collect and store your specimens for future research, the following will happen:

Blood, Saliva, and/or Cheek Cell Sample Donation

- Approximately four teaspoons of blood will be drawn for these tests. If you/your child wish to participate in the study but are unable to have a blood test, we would accept a sample of saliva in a specified collecting kit. We may also collect a cheek swab sample in a specified collecting kit. DNA will be extracted and cell lines will be generated from the white blood cells to study the causes of disorders of brain development. These DNA samples and cell lines will be coded and may be shared with collaborating investigators to study brain abnormalities.
- If you/your child visit us at UCSF or are able to meet our research team at a conference, we will arrange to have your/your child's blood drawn by our clinic or by a contracted nurse at the conference.
- If you/your child are unable to visit us, we may provide you with a kit for collection of specimen and shipping materials for sending it back. You can take this kit to your local doctor or laboratory and have the blood samples drawn there. If you are unable to donate blood samples, we will mail you a saliva collection kit and/or cheek cell swab kit that will contain all of the supplies necessary to collect your saliva and/or cheek cell sample and have it sent back to us.
- The blood samples will be kept in storage in what is called a "tissue bank". We do not know for sure if your/your child's specimen will be used, our plan is to use it for research about brain disorders.
- We will also ask you to provide information to us for review including (but not limited to) your/your child's medical and school records, neuropsychological testing reports and therapy evaluations. We will also ask you/your child to undergo a physical examination, provide information about your/your child's medical history and picture will be taken.
- We will continue to gather additional information from you/your child annually and this will continue until the end of the study.

Hair donation

- If you/your child are interested in donating a hair sample, we will ask you/your child to pluck several hairs from your scalp and place them in a container provided to you. Analysis of hair samples may be able to teach us important information that cannot be learned from studying blood alone.

Skin sample from research procedure donation

- A small group of interested participants will take part in a research biopsy, and the specifics of the procedure will be covered in a separate consent form. If you are interested in having a skin biopsy, on yourself or your child, done through our research program, please contact the research staff, and the research staff will provide you with additional information.

Skin sample from medical procedure donation

- We are also interested in studying tissue obtained during medical procedures, for example a skin or muscle biopsy. Analysis of this tissue can often teach us important information about diseases that cannot be learned from studying blood alone. If you or your child will be having a tissue biopsy for medical purposes, and only for medical purposes, and you are interested in donating part of this tissue, please inform one of the members of the research staff and they will provide additional information.
- After we receive notification from you about a medical procedure, we will ask you to provide some basic information including:
 1. What type of medical procedure will be performed? Will it be a biopsy, surgery, amniocentesis, pregnancy termination, autopsy, etc.
 2. Date and time of scheduled procedure.
 3. Name of physician performing procedure and their contact information.
- We will review this consent form with you and be available to answer any questions.
- We will contact your/your child's physician and provide her/him with a copy of this consent
- Dr. Sherr or a member of his research team will ask the physician to collect a sample of tissue for the study. After all routine tests required for your/your child's care are finished, instead of discarding your leftover specimen, your treating physician will be asked to save it and send it to our study for storage in what is called a "tissue bank". We do not know for sure if your specimen will be used, but they might be used in research about brain disorders.
- If any additional tissue (including amniotic fluid) will be taken for research purposes, a sliver of tissue (the size of a grain of rice) from the same site that the medical procedure is being performed will be requested or less than one teaspoon of additional amniotic fluid. The physician will be asked not to collect extra tissue or fluid if she/he determines that it will cause any additional risk, discomfort, or pain beyond what is normally expected from the procedure.

- Cells obtained from a skin biopsy will be grown in the laboratory so that specialized chromosome tests can be done. In addition, these cells will be induced to differentiate into pluripotent stem cell.

Genetic information (also known as genotype data) and the medical record data (also known as phenotype data) may be shared broadly in a coded form for future genetic research or analysis. We may give the specimen and certain medical information about you/your child (for example, diagnosis, age, seizure history, etc.) to other scientists not at UCSF including to a controlled access government health research database, but we will not give them your/your child's name, address, phone number, or any other information that would identify you/your child. Data may only be used for research on the specific neurological disorders relevant to this study. Reports about any research will not be given to you or your/your child's doctor. Sometimes specimens are used for genetic research (about diseases that are passed on in families). Even if we use the specimen for genetic research, we will not put the results in your/your child's medical record. The research will not change the care you/your child receive(s). Your/your child's specimen and any information about you/your child will be kept until it is used up or destroyed. It may be used to develop new drugs, tests, treatments or products. In some instances these may have potential commercial value. Your/your child's personal health information cannot be used for additional research without additional approval from either you or a review committee.

Your/your child's specimen will be kept indefinitely. If you decide later that you do not want your/your child's sample and information to be used for future research, you can tell us, and we will destroy any remaining identifiable sample and information if it is no longer needed for your/your child's care. However, if any research has already been done using portions of your specimens, the data will be kept and analyzed as part of those research studies.

What risks are involved with donating specimens for research?

- Venipuncture: The risks of drawing blood include temporary discomfort from the needle stick and bruising.
- Any extra tissue or amniotic fluid your physician takes from you or your child will come through cuts or tubes needed for your medical procedure, and should not cause you any risk, discomfort, or pain beyond what we normally expect from surgery.
- Confidentiality: Donating specimens may involve a loss of privacy, but information about you will be handled as confidentially as possible. Your/your child's name will not be used in any published reports from research performed using your specimen. The manager of the tissue bank (Dr. Elliott Sherr) and select tissue bank staff members will have access to information about you/your child but they will not release any identifying information about you/your child to researchers using your/your child's specimen. The UCSF Committee on Human Research and other University of California personnel also may see information about you/your child to check on the tissue bank. Once your/your child's health information is disclosed to the research team it is not protected under the Health Information Portability and Accountability Act (HIPAA). The tissue bank staff will continue to protect your/your child's personally identifiable health information as described in this consent form.

The University of California complies with the requirements of HIPAA and its privacy regulations, and with all other applicable laws that protect the confidentiality of your/your child's health information.

- 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.
- There is a risk that someone could trace your information and sample back to you. Even without your name or other identifiers, your genetic information is unique to you. The researchers believe the chance that someone will identify you is very small, but the risk may change in the future as people come up with new ways of tracing information.
- Donating and broad sharing of data may involve a loss of privacy, but information about you will be handled as confidentially as possible. Study data will be physically and electronically secured. As with any use of electronic means to store data, there is a risk of breach of data security. Genetic information that results from this study does not have medical or treatment importance at this time. However, there is a risk that information about taking part in a genetic study may influence insurance companies and/or employers regarding your health. Taking part in a genetic study may also have a negative impact or unintended consequences on family or other relationships. It is possible that future research could one day help people of the same race, ethnicity, or sex as you. However, it is also possible through these kinds of studies that genetic traits might come to be associated with your group. In some cases, this could reinforce harmful stereotypes. In some cases, this information could be used to make it harder for you to get or keep a job or insurance. There are laws against the misuse of genetic information, but they may not give full protection. There can also be a risk in knowing genetic information. New health information about inherited traits that might affect you or your blood relatives could be found during a study. The researchers believe the chance these things will happen is very small, but cannot promise that they will not occur.

What happens if I am injured because I took part in this study?

It is important that you tell your study doctor, Dr. Elliott Sherr, if you feel that you/your child have been injured because of taking part in this study. You can tell the doctor in person or call him at 415-502-8039.

Treatment and Compensation for Injury: If you are injured as a result of being in this study, the University of California will provide necessary medical treatment. The costs of the treatment may be billed to you or your insurer just like any other medical costs, or covered by the University of California, depending on a number of factors. The University and the study sponsor do 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 the benefits of donating specimens for research?

There will be no direct benefit to you/your child for allowing your/your child's specimen to be used for research. Research results will not be returned to you/your children. However, we hope we will learn something that will help in the treatment of future patients through the advancement of science and

understanding of health and disease.

What financial issues should I consider before donating?

You/your child will not be charged for donating your/your child's specimen. You/your child will not be paid for donating your/your child's specimen. If any new products, tests or discoveries that result from this research have potential commercial value, you/your child will not share in any financial benefits. UCSF may receive payment from researchers requesting specimens in order to cover the costs of collecting and storing the specimens.

What alternatives do I have?

You can choose not to donate your/your child's specimen and/or you/your child can choose not to be a part of the overall study on disorders of cerebral development.

What are my rights if I take part in this study?

Taking part in this study is your choice. You/your child may choose either to take part or not to take part in the study. No matter what decision you make, there will be no penalty to you/your child. In the case of injury resulting from this study, you/your child do(es) not lose any of your/their legal rights to seek payment by signing this form.

Can I stop being in the study?

If you first agree to let you/your child participate and then change your mind, you are free to withdraw yourself/your child from the study and discontinue your/your child's participation at any time. To do so, you can notify the investigator in writing at the address below and we will destroy any remaining data. However, if any research has already been done using portions of your child's specimens or distributed to other researchers, the data cannot be retracted. 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.

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Who can answer my questions about the study?

You can talk to the researchers about any questions or concerns you have about this study. Contact the researcher(s), Dr. Elliott Sherr at 415-502-8039.

For questions about your rights while taking part in this study, call the office of the **Committee on Human Research**, UCSF's Institutional Review Board (a group of people who review the research to protect your rights) at **415-476-1814**.

Consent

Please read each sentence below and think about your choice. If you have any questions, please talk to Dr. Elliott Sherr, or call our research review board at IRB's phone number.

No matter what you decide to do, it will not affect your care. After reading each sentence, put your INITIALS in the "Yes" or "No" box

YES	NO
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1. My/my child's tissue may be kept for use in research to learn about, prevent, or treat brain disorders.

YES	NO
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2. My/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).

YES	NO
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3. Additional tissue may be taken for this research, as described in the What Will Happen If I Agree... section above.

You/your child 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/your child are/is otherwise entitled.

You will be asked to sign a separate form authorizing access, use, creation, or disclosure of health information about you/r child. PARTICIPATION IN RESEARCH IS VOLUNTARY. You will be given copies of this consent form and the Experimental Subject's Bill of Rights to keep.

When subject is an adult:

Please note that all individuals providing blood samples and/or medical records are considered subjects. Therefore, all participating relatives must sign a copy of this form.

Date	Subject's Signature for Consent	Print Name of Adult Subject
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If the person being considered for this study is unable to consent himself/herself because he or she is cognitively impaired the legally authorized parent or legal guardian my sign here:

Date	Parent or Legal Guardian's Signature	Print Name
		Relationship to Adult Subject

When subject is a minor:

If the subject being considered for the study is a minor, the parents or legal guardians may sign here:

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

If your child is able to read and understand the procedures, risks, and benefits of the study, please have them indicate their willingness to participate by signing below, in addition to your signature above.

Date	Signature of Minor	Print Name
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Date

Signature of Person Obtaining Consent

Print Name of Person Obtaining Consent