

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

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 characterized with social and/or behavioral deficits, or is suspected of developmental delay.

AND/OR

2. He or she has a particular chromosomal change. A chromosomal change is a specific change inside the DNA of a person that may affect their development and/or health.

Why is this study being done?

Dr. Sherr and his colleagues at UCSF conduct clinical research seeking to advance the understanding of developmental disorders. With these investigations, we are trying to understand how biochemical pathways may affect thinking, behavior and overall development.

How many people will take part in this study?

Approximately 100 people with developmental disorders or chromosomal change will take part. Additionally, 200 parents and 50 siblings will take also enroll and take part.

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 may 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 Pediatric Clinical Research Center (PCRC) or the UCSF Neurosciences Clinical Research Unit (NCRU) at Mission Bay 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 five tubes of blood (10-30mL, approximately one to two tablespoons) 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. You and/or your child may be asked if they would like numbing cream (LMK) to reduce the discomfort associated with the blood draw. The research coordinator will administer the cream 20-30 minutes before the blood draw.
4. You and/or your child may be asked to provide an additional blood sample for further analysis at a later study visit. Approximately 10-20 mL of blood will be collected during each visit for up to two additional visits. The additional samples will not be obtained in the same visit as the first blood donation but would occur at least two weeks apart for each draw. The volume of blood that will be drawn in this study will not exceed 50 mL over an 8 week period. Giving an additional sample of blood is entirely optional.
5. Your child's height, weight, and head size will be measured. Vitals will also be taken.
6. Your child may be asked to complete an evaluation with a psychologist at the UCSF Pediatric Clinical Research Center (PCRC). 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.
7. You and/or your child may also be asked if you and/or your child would allow the in-person cognitive testing session to be videotaped. Videotaping is used to ensure research reliability in testing administration and accurate scoring of the testing session.

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). To reduce the discomfort from the needle stick, your child may be asked if they would like to use numbing cream.

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.

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 certain medical information about you (for example, diagnosis, blood pressure, age if less than 85) to other scientists or companies not at UCSF, including to a controlled access government health research database, but we will not give them your name, address, phone number, or any other identifiable information. Research results from these studies will not be returned to you.

Donating 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.

There will be no direct benefit to you from allowing your data to be kept and used for future research. However, we hope we will learn something that will contribute to the advancement of science and understanding of health and disease. If the data or any new products, tests or discoveries that result from this research have potential commercial value, you will not share in any financial benefits. If you decide later that you do not want your information to be used for future research, you can notify the investigator in writing at (415)502-8039, and any remaining data will be destroyed. However, we cannot retract any data has been shared with other researchers.

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 your child from participating in this study. However, the information that you and your child provide may help health professionals better understand social deficits observed in developmental disorders and/or other neurologic, psychiatric, or behavioral disorders and 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 developmental disorders 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.

Videotapes

There is a possibility that we may want to use the videotape from your child's neuropsychological exam for training and reliability purposes only. The videotape will

include full facial features and voice recording. These clinicians will not receive information about your child's last names, addresses, or dates of birth. If you agree to this, the videotape may be viewed by clinicians who conduct this research study at different universities. These videotapes will be stored in locked cabinets and will be kept indefinitely. Your decision regarding outside use of your videotape will not affect your family's participation in this research study. You will not receive any additional compensation for these tapes.

Please write your INITIALS on the appropriate line below to indicate your preference.

____ I allow the videotape of the neuropsychological exam. It will be used for training, reliability and additional analysis purposes.

____ I do NOT allow the videotape of my child's neuropsychological exam.

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, you will receive compensation in cash or check upon completion of all of the study activities described under the section, "What will happen if I take part in this research study?" You may also be given a prepaid debit card for taking part in this study. We will give you separate instructions on how to use the debit card.

Your child will receive \$20 for each blood draw and height, weight, and head size measurements.

Your child will receive \$30 for the completion of all surveys and questionnaires

Your child will receive \$30 for completion of the in-person psychological assessments.

In the event that you are reconsenting for the purpose of providing data linkage to this study, and the study components listed above have already been completed, there will be no additional study compensation provided.

If completion of the study components listed above is satisfied by data linkage, no study visit is required to collect that data or sample and no study compensation will be provided for that study component.

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.

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). 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). If any new products, tests or discoveries that result from this research have potential commercial value, you and your child will not share in any financial benefits.
- 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.
- 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.
- 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.

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 and/or behavioral disorders.
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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. 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.
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OPTIONAL: Affiliated Study Data and Sample Linkage

If you have participated in other studies affiliated with the leading investigator of this study (Dr. Elliott Sherr) OR with the sponsor of the study (e.g. the Simons Foundation, Simons Simplex Collection etc.), there may be opportunities to use previously collected information or

biological samples from these prior studies to contribute to this current project. We are also asking for your consent to share the data we collect during this study here at the University of California, San Francisco with the Simons Foundation and the SSC@IAN in order to add to the information that was collected during your participation in the SSC. Please note your decision to indicate your participation in other studies is **OPTIONAL** and **VOLUNTARY**. **After reading each sentence, please initial either the "Yes" or "No" box**

YES	NO
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I have participated in other studies affiliated with the leading investigator of this study (Dr. Elliott Sherr) OR with the sponsor of the study (Simons Foundation, Simons Simplex Collection).

Name of Study: _____

YES	NO
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I allow the research team to **obtain** and use my information/biological samples, previously collected from other studies affiliated with the leading investigator of this study (Dr. Elliott Sherr) OR sponsor of the study (Simons Foundation, Simons Simplex Collection)

YES	NO
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I allow the research team to **share** information obtained about me during this study to be made available to the Simons Foundation. Data that will be shared may include, identifying information, demographic information, survey measures, medical history, body measurements, and data derived from the collected blood sample.

Please note that it is not required for your participation to indicate whether or not you participated in another study.

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