

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

### **Study Title: Brain Imaging and Cell Signaling: Insights into the Biology of Autism**

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 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.

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

1. Your child has been given a diagnosis of Autism Spectrum Disorder (ASD), a similar disorder characterized by social and/or behavioral deficits, or is suspected of developmental delay.

AND/OR

2. Has a head circumference that is characterized as macrocephalic, normalcephalic, or microcephalic.

AND/OR

3. Your child has a particular chromosomal or genetic change. A chromosomal/genetic 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 autism spectrum disorders (ASD) as well as other neurodevelopmental disorders. With these investigations, we are trying to understand how biochemical pathways, brain anatomy, and brain connectivity may affect thinking, behavior and overall development.

### **How many people will take part in this study?**

Approximately 50 people with ASD and macrocephaly or other developmental disorders or genetic changes will take part, as well as 50 matched macrocephalic controls. Approximately 40 people with ASD and normalcephaly or other developmental disorders or genetic changes will take part, as well as 40 matched normalcephalic controls. Approximately 50 people with ASD and microcephaly or other developmental disorders or genetic changes will take part, as well as 50 matched controls. Additionally, 150 parents and approximately 100 siblings will

also enroll. Of the 140 ASD individuals, we anticipate approximately 100 individuals will be eligible for the imaging portion of the study.

### **What will happen if I take part in this research study?**

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

1. The researchers will gather information about you from a brief telephone interview or online questionnaire.
2. The researchers will also measure the head circumference of your child and the head circumference of both parents. This can be done at home with our guidance over the phone or in person.
3. You will visit the Pediatric Clinical Research Center (PCRC) or the UCSF Neurosciences Clinical Research Unit (NCRU) at Mission Bay and you will give a blood sample. The blood will be drawn by putting a needle into a vein in your arm. One to five tubes of blood (10-30mL, approximately one to two tablespoons) will be taken. This will take about five minutes.
4. You may be asked if you would like numbing cream to reduce the discomfort associated with the blood draw. The research coordinator will administer the cream 20-30 minutes before the blood draw.
5. You 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.
6. Your height, weight, and head size will be measured. Vitals will also be taken.
7. Your child may be asked to complete an evaluation with a psychologist and/or a research team member 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 researcher may ask you to be present in the room during the test.
8. You will be asked to have a close informant complete a series of standardized survey measures about your social, behavioral, and developmental traits. We will ask the surveys to be completed by all enrolled family members. The surveys will take about 1 hour to complete, and can be done at home or over the phone with the research staff.

### **How long will I 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 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?**

#### **Blood Draw**

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

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. This study may help us to understand better behavioral, cognitive, and structural and functional brain anatomy in autism spectrum disorders and/or other neurologic, psychiatric, or behavioral disorders and may support the development of novel therapeutic strategies.

#### **Blood Sampling:**

Results of the blood sampling will not be shared with participants as they are for research purposes only.

### **What other choices do I have if my child does 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 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 you. Information that 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 that identifies you will be destroyed when this research is complete. Your personal information may be given out if required by law. The following organizations may look at information about you in your medical and research records:

- Study Sponsor: Simons Foundation
- 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, 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 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 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. We will reimburse you for parking costs if you visit UCSF to complete any of the research activities described under the section, "What will happen if my I take part in this research study?"

### **Will I 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.

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

You will receive \$30 for the completion of all surveys and questionnaires.

### **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 their regular benefits. Leaving the study will not affect your medical care. You 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 you 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.

### **What will happen if I agree to donate my specimens?**

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 You's 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). Even if we use your tissue for genetic research, we will not put the results in your medical record. The research will not change the care you receive. Your specimen and any information about you 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 personal health information cannot be used for additional research without additional approval from either you or a review committee.
- We may give your tissue specimen(s) and certain medical information about your (for example, diagnosis, age, developmental history, etc.) to other scientists or companies not at UCSF, including a controlled access government health research database or the study sponsor (Simons Foundation), 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). If any new products, tests or discoveries that result from this research have potential commercial value, you will not share in any financial benefits.
- Your 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 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.

## Risks

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 You's care.

YES	NO	1. My 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 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 with the sponsor of the study (Simons Foundation, Simons Simplex Collection).

YES	NO
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I allow the research team to **share** information/biological samples 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 they participated in another study.**

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## 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

By signing below, you are giving your permission to be included in this study.

_____	_____	_____
Date	Signature	Print Name

### 2. Study Staff Signature

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

### 3. Legally Authorized Representative Consent

If you are the Legally Authorized Representative, the person you are representing (hereafter referred to as the 'subject') is being asked to participate in a research study but is unable to consider whether to give consent to participate because of their medical condition. You, as the subject's legally accepted representative, are being asked to consider whether to give consent for the subject to participate in this study.

Date	Legally Authorized Representative
Date	Person Obtaining Consent

### 4. Subject Re-consent

I have been enrolled in this study by a surrogate because my illness rendered me incapable of providing informed consent when I was initially enrolled. I have since regained my ability to provide consent and now would like to:

Continue participation until the study is complete

Terminate my participation in the study but allow the use of data collected to date

Terminate my participation in the study and not allow the use of data collected to date

Date	Participant's Signature for Consent
Date	Person Obtaining Consent