A. PURPOSE AND BACKGROUND
You are currently participating in a research study being conducted by Dr. Elliott H. Sherr and his colleagues to learn about brain development and disorders that can occur in this process. You have already signed the main consent form and have indicated your participation in the main research study. You are now being asked to provide consent for a skin biopsy procedure. The tissue obtained from your skin will allow the researchers to use additional approaches to discover genetic causes of disorders of brain development. Participation in this study is completely voluntary. The test is safe and can be performed using local anesthetic. The individual with a disorder of brain development will be invited to participate in the skin biopsy study. Only those subjects who participate in the main study are eligible to participate in the skin biopsy portion of the study. Approximately 20 participants will be involved with this component of the study.

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. 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.

Please read the information below and ask questions about anything that you do not understand before deciding if you want to participate. The researchers will be available to answer your questions. In order to decide whether or not you want to be part of this research study, you should know enough about its risks and benefits to make an informed judgment. This consent form gives detailed information about the skin biopsy that will be discussed with you. You will be asked to sign this form if you wish to participate.

B. PROCEDURE:
The area of skin will be numbed with lidocaine injection. Skin is usually taken from the forearm. After 5 minutes the skin area will become numb. The numb skin area will be cleaned with alcohol and a 3 mm skin sample the size of a pencil eraser imprint will be obtained using a standard punch biopsy needle. The sample of skin will be snipped at the base with a sterile pair of scissors and placed in a tube containing a fluid that allows the cells to grow. You may feel some discomfort when the skin sample is taken. The total amount of time for the skin biopsy procedure is approximately 10 minutes for the actual procedure to be performed. No stitches are required. Bacitracin ointment will be applied to the biopsy site. The biopsy site should have a clean application of bacitracin ointment and a clean bandage applied daily for three days.

The sample will be placed in a specimen bag and delivered to our lab at UCSF. The skin sample will be cultured (which means the skin cells are fed with nutrients in the lab to grow a certain type of cell called fibroblast cells). This lets us have a large supply of cells that can be used for
research. The cultured cells will allow us to do specialized chromosome tests. In addition, these cells will be induced to differentiate into pluripotent stem cells as explained above. All cells will be stored at the lab with your consent and will be available to Dr. Sherr for his research. All identifiable information that will be collected about you will be removed from the sample and replaced with a code.

You may at any time contact the researchers at 415-502-8039 and ask that your samples be withdrawn from research use, and any identifiable samples still in their possession will be destroyed.

C. RISKS AND DISCOMFORTS:
The possible risks and/or discomforts associated with the procedures described in this study are minimal or/and moderate. These are as follows:

At the time of collection of a 3 mm sample of skin, there will be some discomfort or pain from the skin biopsy punch insertion, and there is some possibility of bruising, swelling, bleeding, and infection at the site of the scalpel punch insertion, also rarely of fainting. Please inform Dr. Sherr and your local physician if you experience pain or bleeding from this skin biopsy procedure. No stitches are required. The resulting scar from the biopsy is tiny and does not cause problems. There may be some discomforts associated with the lidocaine injection.

There is a reasonable possibility that no findings will result from this research effort. Any significant findings that do result may take months or years to complete. If you wish to inquire into the progress of our research, you are welcome to do so at any time.

Confidentiality:
You have been asked to sign a separate form authorizing access, use, creation, or disclosure of health information about you. All identifiable information that will be collected about you will be removed and replaced with a code. A list linking the code and your identifiable information will be kept separate from the research data. All research data will be maintained in a secure location at UCSF. Only authorized individuals will have access to it.

Treatment and Compensation for Injury:
If you are injured as a result of being in this study, treatment will be available. The cost of such 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.

D. 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.

E. BENEFITS:
There will be no direct benefit to you (other than the data we mentioned above) from participating in this study. However, it is hoped that the information gained from the study will help in the treatment of future patients with conditions like this and will help the researchers learn more about how the structure of the brain is formed.

F. ALTERNATIVES:
Participation in the research is completely voluntary. You should not feel any pressure to participate. If you do not want to participate it will not interfere with any current or future care you or your family receives at this institution. Participants who choose not to participate in this skin biopsy portion of the study can still participate in the main study.

G. COSTS:
All procedures related to the skin biopsy will be provided at no cost to you.

H. PAYMENT
You will not be paid for your participation in this study.

I. QUESTIONS
This study has been explained to you by Dr. Elliott Sherr or the person who signed below and your questions were answered. If you have any other questions about the study, you may call Dr. Sherr at (415) 514-9306 or his associate at (415) 502-8039.

J. CONSENT
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 from this study. You will be given copies of this consent form and the Experimental Subject’s Bill of Rights to keep.

If you would like to participate, you should sign below.

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<tr>
<th>Date</th>
<th>Subject’s Signature for Consent</th>
<th>Print Name</th>
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Skin Biopsy Consent Adult          Page 3 of 4          Modification Date: 04/29/11
If the subject 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 may sign here:

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