If you are using Epic for this study, fax a copy of the signed consent form to 410-367-7382.

Patient I.D. Plate

PARENT INFORMED CONSENT AND PRIVACY AUTHORIZATION FORM

Protocol Title: Sleep dysfunction in children with pathogenic SYNGAP1 mutations

Application No.: IRB00183481

Principal Investigator: Dr. Shilpa Kadam
707 N. Broadway Baltimore, MD 21205
Work: 614-440-3811
Fax: 443-923-2695

1. What you should know about this study:

- You are being asked to allow your child to join a research study. This consent form explains the research study and your child's part in the study. Please read it carefully and take as much time as you need. Ask your study doctor or the study team to explain any words or information that you do not understand.
- Joining this study is voluntary. If you allow your child data to be included in the study, you can change your mind later. There will be no penalty or loss of benefits if you decide not to allow your child's de-identified data to be part of the study.
- During the study, we will tell you if we learn any new information that might affect whether you wish to continue to allow your child to participate.
- If we think your child's participation in this study may affect your child's clinical care, information about your child's study participation will be included in your child's medical record, which is used throughout Johns Hopkins. Doctors outside of Johns Hopkins may not have access to this information. You can ask the research team to send this information to any of your child's doctors.
- When Johns Hopkins is used in this consent form, it includes The Johns Hopkins University, The Johns Hopkins Hospital, Johns Hopkins Bayview Medical Center, Howard County General Hospital, Johns Hopkins Community Physicians, Suburban Hospital, Sibley Memorial Hospital and All Children's Hospital.

2. Why is this research being done?

This research is being done to better understand the relationship between SYNGAP1 mutations, epilepsy, and sleep dysfunction.

Patients with SYNGAP1 mutations often report sleep dysfunction and seizures on their electroencephalograms (EEGs). We plan to analyze previously recorded overnight/24 hour EEGs to see if we can learn more about the relationship between these things.

Children aged 2-16 years with SYNGAP mutations, who have previously recorded overnight/24h electroencephalograms (EEG) or polysomnogram studies, may join.

How many children will be in this study?
Approximately 30 children are expected to take part in this study.

3. What will happen if you allow your child to join this study?

This is not a treatment study. This study does not involve any procedures or patient interaction.

If you agree to allow your child to be in this study, we will ask you to allow us to do the following things:

- Allow our study group to analyze child's clinical history, medical history, and a previously recorded overnight EEG.

4. What are the risks or discomforts of the study?

Your child will not experience any physiological risks or discomforts as this study does not involve any procedures or patient interaction.

Confidentiality
There is the risk that information about you or your child may become known to people outside this study.

The overnight EEG, clinical history, and medical history usually have child's identity stamped on the data that you will voluntarily release to the PI. However, to prevent this during the process of analyses all data will be de-identified by the PI before proceeding with any further analyses. All raw data CDs and associated reports will be locked in a file closet with only PI access. All de-identified electronic data files will stored on the KKI intranet in login-only accessible folders that only the PI and authorized staff will have. Access to files will be given to authorized personnel only.

5. Are there benefits to your child from being in the study?

There is no direct benefit to your child from being in this study. If your child takes part in this study , your child may help others in the future.

6. What are your options if you do not want your child to be in the study?

You do not have to allow your child to join this study.

If your child does not take part in the study, your child's care at Kennedy Krieger will not be affected.

7. Will it cost you anything to allow your child to be in this study?

No.

8. Will you or your child be paid if you allow your child to join this study?

No.

9. How will your child's privacy be protected?

We have rules to protect information about your child. Federal and state laws and the federal medical Privacy Rule also protect your child's privacy. By signing this form you provide your permission, called your "authorization," for the use and disclosure of information protected by the Privacy Rule.

The research team working on the study will collect information about your child. This includes things learned from the procedures described in this consent form. They may also collect other information including your child's name, address, date of birth, and information from your child's medical records

(which may include information about HIV status, drug, alcohol or STD treatment, genetic test results, or mental health treatment).

The research team will know your child's identity and that your child is in the research study. Other people at Johns Hopkins, particularly your doctors, may also see or give out your child's information. We make this information available to your child's doctors for your child's safety.

People outside of Kennedy Krieger may need to see or receive your child's information for this study. Examples include government agencies (such as the Food and Drug Administration), safety monitors, other sites in the study and companies that sponsor the study.

If your child is in a cancer study that receives federal funding, the National Cancer Institute (NCI) now requires that we report identifiable information (such as, zip code) about your child's participation. You may contact the NCI if you have questions about how this information is used.

We cannot do this study without your authorization to use and give out your child's information. You do not have to give us this authorization. If you do not, then you may not join this study.

We will use and disclose your child's information only as described in this form and in our Notice of Privacy Practices; however, people outside Kennedy Krieger who receive your child's information may not be covered by this promise or by the federal Privacy Rule. We try to make sure that everyone who needs to see your child's information keeps it confidential – but we cannot guarantee that your information will not be re-disclosed.

The use and disclosure of your child's information has no time limit. You may revoke (cancel) your permission to use and disclose your child's information at any time by notifying the Principal Investigator of this study by phone or in writing. If you contact the Principal Investigator by phone, you must follow-up with a written request that includes the study number and your contact information. The Principal Investigator's name, address, phone and fax information are on page one of this consent form.

If you do cancel your authorization to use and disclose your child's information, your child's part in this study will end and no further information about your child will be collected. Your revocation (cancellation) would not affect information already collected in the study, or information we disclosed before you wrote to the Principal Investigator to cancel your authorization.

10. Will the study require any of your other health care providers to share your child's health information with the researchers of this study?

As a part of this study, the researchers will require to see your child's health care records from her/his other health care providers. Therefore, you will be asked to give us a list of other health care providers that your child uses.

11. What other things should you know about this research study?

a. What is the Institutional Review Board (IRB) and how does it protect you?

The Johns Hopkins Medicine IRB is made up of:

- Doctors
- Nurses
- Ethicists
- Non-scientists
- and people from the local community.

The IRB reviews human research studies. It protects the rights and welfare of the people taking part in those studies. You may contact the IRB if you have questions about your child's rights as a participant or if you think you or your child have not been treated fairly. The IRB office number is 410-955-3008. You may also call this number for other questions, concerns or complaints about the research.

When the Johns Hopkins School of Medicine Institutional Review Board (IRB) reviews a study at another site, that site (institution) is solely responsible for the safe conduct of the study and for following the protocol approved by the Johns Hopkins IRB.

b. What do you do if you have questions about the study?
Call the principal investigator, Dr. Shilpa Kadam at 443-923-2688. If you wish, you may contact the principal investigator by letter or by fax. The address and fax number are on page one of this consent form. If you cannot reach the principal investigator or wish to talk to someone else, call the IRB office at 410-955-3008.

c. What happens to Data that are collected in the study?
Kennedy Krieger and our research partners work to understand and cure diseases. The data you provide are important to this effort.

If you allow your child to join this study, you should understand that you/your child will not own your child's data, and should researchers use them to create a new product or idea, you/your child will not benefit financially.

12. Assent Statement

This research study has been explained to my child in my presence in language my child can understand. He/she has been encouraged to ask questions about the study now and at any time in the future

13. What does your signature on this consent form mean?

Your signature on this form means that:

- you understand the information given to you in this form
- you accept the provisions in the form
- you agree to allow your child to join the study

You and your child will not give up any legal rights by signing this consent form.

WE WILL GIVE YOU A COPY OF THIS SIGNED AND DATED CONSENT FORM

_____

Signature of Parent (Print Name) Date/Time

_____

Signature of Person Obtaining Consent (Print Name) Date/Time

_____

Signature of Legally Authorized Representative (LAR) (Print Name) Date/Time
For CHILD PARTICIPANT

_____

Description of LAR's authority under Maryland Law to act as surrogate health care decision-maker for child research participant (for example, Legal Guardian; Court-ordered representative) Date/Time

NOTE: A COPY OF THE SIGNED, DATED CONSENT FORM MUST BE KEPT BY THE PRINCIPAL INVESTIGATOR; A COPY MUST BE GIVEN TO THE PARTICIPANT; IF YOU ARE USING EPIC FOR THIS STUDY A COPY MUST BE FAXED TO 410-367-7382; IF YOU ARE NOT USING EPIC A COPY MUST BE PLACED IN THE PARTICIPANT'S MEDICAL RECORD (UNLESS NO MEDICAL RECORD EXISTS OR WILL BE CREATED).

ONLY CONSENT FORMS THAT INCLUDE THE JOHNS HOPKINS MEDICINE LOGO CAN BE USED TO OBTAIN THE CONSENT OF RESEARCH PARTICIPANTS.