INFORMATION SHEET FOR CAREGIVERS OF INDIVIDUALS WITH NEURODEVELOPMENTAL DISORDERS

SYNGAP1 and RAI1 Deficiencies, and Fragile X Syndrome (FXS)

Study Title:
Development of Caregiver Reported Outcome Measures for Neurodevelopmental Disorders

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This information sheet describes a research study that is being conducted by Dr. Chad Heatwole, MD, MS-CI, from the University of Rochester Department of Neurology in Rochester, New York, United States.

The purpose of this study is to develop and validate a disease-specific, observer-reported outcome measure for clinical trials of patients with the following Neurodevelopmental Disorders (NDDs): SYNGAP1 and RAI1 deficiencies, and Fragile X Syndrome. The study is being conducted in multiple phases. We are contacting you regarding Phase 1, which involves interviews with caregivers of patients with SYNGAP1 and RAI1 deficiencies, and Fragile X Syndrome. We estimate that approximately 75 caregivers, ages 18 and up, who care for an individual with NDDs will take part in Phase 1.

As part of Phase 1, if you agree to participate, you will complete a phone interview that includes providing background and demographic information of you and the person with an NDD for whom you care and identifying important symptoms for patients with NDDs that affect the life of the person with an NDD for whom you care. We anticipate that the phone interview will take one hour or less. The interview will be audio-recorded and transcribed, and the data analyzed.

There is a minimal risk associated with this research. Because this study involves collecting personal information about you, there is a potential for invasion of privacy or breach in confidentiality. To minimize this risk, all interview recordings and transcripts will be kept in a secure, locked location. Names or other identifiable information will not be included on the recordings or transcripts. The questions that you will be asked to answer during the interview are personal and may make you feel uncomfortable or upset. You may decline to answer any questions that you do not want to answer. You will also be given the option to withdraw from the interview at any time.

You and the person you whom you care will not receive direct benefits from participating in this study. The data from the interviews will assist in the development of an observer-reported outcome instrument for patients with NDDs, to be used in clinical trials and patient monitoring. This instrument will, ultimately, allow future therapies in NDDs to be directly evaluated based on symptoms and domains that you (and others) identify as important.

The University of Rochester is receiving funding for conducting this research. You will not be paid for participating in this study. There are no expected costs to participating in this study.

Authorization to Use Health Information: The University of Rochester makes every effort to keep the information collected from you private. Therefore, your interview will be recorded anonymously. We will store and transcribe the recorded conversation in a secure manner, and only study team members will have access to it. Transcripts will be stored indefinitely. Sometimes, however, researchers need to share information that may identify you with people that work for the University, regulators, or the study sponsor. If this does happen, we will take precautions to protect the information you have provided. Results of the research may be presented at meetings or in publications, but your name and name of the person you care for will not be used.

In order to collect study information, we have to get your permission to use and store your personal health information. The audio-recorded interview that contains your voice is considered one of the HIPAA identifiers. Recordings will be kept in a locked, secure location for up to five years after this research ends. Your name and other personally-identifiable information will NOT be included in these recordings; voice recording is the only HIPAA identifier that we require your permission to use. By law, we have to get your authorization to use health information we collect from you about the person you care for. We may use your direct insights and quotes or research observations made while you take part in this research. This information may be audited to ensure we are following regulations, policies, and study plans. Your permission to use the health information you provide for this study will not expire unless you tell us you want to cancel it. We will keep the information we collect about you and the person you care for indefinitely. If you cancel your permission, you will be removed from the study.

Use of Email in Research: Email communications may be sent or received in an unencrypted (unprotected) manner. Therefore, there is a risk that the content of the communication, including your personal information, could be shared beyond you and the study team. Your consent in participating in this research study indicates that you understand this risk. The University of Rochester is not responsible for any interception of messages sent through email. E-mail can be intercepted, altered, forwarded, or used without authorization or detection. E-mail can be used to introduce viruses into computer systems.

Participation in this study is voluntary: You are free not to participate or withdraw at any time, for whatever reason without penalty or loss of benefit to which you are entitled. In the event that you do withdraw from this study, the information you have already provided will be kept in a secure manner.

For more information or concerns regarding this research, please contact
Jennifer Weinstein, email jennifer.weinstein@chet.rochester.edu
Jamison Seabury, email jamison.seabury@chet.rochester.edu
Charlotte Engebrecht, email charlotte.engebrecht@chet.rochester.edu

Please contact the University of Rochester Research Subjects Review Board at 265 Crittenden Blvd, CU 420628, Suite 1-250, Rochester, NY 14642, Telephone (585) 276-0005 or (877) 449-4441 for the following reasons:

- You wish to talk to someone other than the research staff about your rights as a research subject;
- To voice concerns about the research;
- To provide input concerning the research process;
- In the event the study staff could not be reached.

Thank you for your interest and your time!