

Sample Consent Document 1: *Natural History Study of the Clinical and Molecular Manifestations of Smith-Magenis Syndrome – pediatric consent*

This consent document was used for the recruitment of pediatric participants for a genome sequencing study. It includes language about returning individual genetic results

Important note: This consent document was developed for the Natural History Study of the Clinical and Molecular Manifestations of Smith-Magenis Syndrome. It is not provided as guidance or as a template promoted by NHGRI, but as a reference to inform investigators and IRBs considering these issues. It is important to tailor consent documents for each individual study.

MEDICAL RECORD	CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY • Adult Patient or • Parent, for Minor Patient
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INSTITUTE:

STUDY NUMBER:

PRINCIPAL INVESTIGATOR:
ADJUNCT PRINCIPAL INVESTIGATOR:

STUDY TITLE:

Continuing Review Approved by the IRB on 08/08/13
Amendment Approved by the IRB on 06/27/14 (F)
Parent Consent for Children

Date Posted to Web: 07/09/14

INTRODUCTION

We invite you to take part in a research study at the National Institutes of Health.

First, we want you to know that:

Taking part in NIH research is entirely voluntary.

You may choose not to take part, or you may withdraw from the study at any time. In either case, you will not lose any benefits to which you are otherwise entitled. However, to receive care at the NIH, you must be taking part in a study or be under evaluation for study participation.

You may receive no benefit from taking part. The research may give us knowledge that may help people in the future.

Second, some people have personal, religious or ethical beliefs that may limit the kinds of medical or research treatments they would want to receive (such as blood transfusions). If you have such beliefs, please discuss them with your NIH doctors or research team before you agree to the study.

Now we will describe this research study. Before you decide to take part, please take as much time as you need to ask any questions and discuss this study with anyone you want or with family, friends, or your personal physician or other health professional.

WHY IS THIS STUDY BEING DONE?

The attached Information Sheet on [Chromosome 17 Deletion](#) explains the study and testing that we are planning to do. Please read the sheet carefully, and ask questions about anything that you do not understand. We want you to understand the study before signing the consent form.

The main goals of the study are:

- To understand medical, behavioral, and learning problems in people with
- To learn more about the region of chromosome 17 that is deleted in
- To learn if specific genetic changes in the 17q21.31 region on chromosome 17 results in specific medical problems.

PATIENT IDENTIFICATION	CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY • Adult Patient or • Parent, for Minor Patient File in Section 4: Protocol Consent (1)
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MEDICAL RECORD**CONTINUATION SHEET for either:**

NIH 2514-1, Consent to Participate in A Clinical Research Study

NIH 2514-2, Minor Patient's Assent to Participate In A Clinical Research Study

STUDY NUMBER:

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- To identify features that must be present to make the diagnosis of SMS.
- To learn about the impact on families of having a child with SMS.

WHAT IS INVOLVED IN THE STUDY?

Your child will be seen by a team of medical specialists for evaluation at _____ over several days. Any tests or evaluations performed as part of this study will be fully explained to you and you are free to refuse any test.

Biological materials obtained for research purposes will be stored in our laboratories during the study. Genetic analysis through DNA testing will be performed to further understand the molecular genetic basis for your child's diagnosis; this may involve sequencing of your/your child's entire DNA to look for changes. (See next section.) A cell line may be developed from the blood/tissue sample your child initially provides to allow us to study your child's DNA in the future without having to take another sample. This lets us do more research on SMS if new techniques become available or as new genes within the SMS deletion region are identified. The samples will be studied for research questions having to do with SMS by researchers associated with this research project. If necessary or helpful, DNA, tissue or fluid specimens from you (your child) will be sent to experts at other centers for analysis. In the future, we may be interested in using your (your child's) samples to pursue other research to understand similar neurobehavioral disorders by sharing samples with other laboratories. If we do so, the sample(s) will be coded without personal identifiers. If there are any risks to you or your family associated with these scientific studies that are not covered in this consent form, we will ask for your consent before such studies are performed.

All laboratory test results given to you will be from an approved clinical diagnostic laboratory. If a finding has important health implications for your child, every effort will be made to have it confirmed by an approved diagnostic laboratory, so we can share the test result with you.

Medical students, graduate students in genetic counseling, and doctors in training may be part of your evaluation. In all cases, students will be directly supervised by professionals certified in Clinical Genetics, Genetic Counseling, or other appropriate fields.

By agreeing to participate in this study, you do not lose any rights that you may have regarding access to and disclosure of your child's records.

It is not possible for a young child or a person with delayed development to fully consent to a research study. Some children can understand a simple explanation of the purpose and procedures for this study. The researcher will help your family decide if this is appropriate for your child. Whenever possible, we will try to explain the study to the participant and obtain his or her permission (Assent) to participate and give a blood sample.

PROVIDING YOU WITH YOUR GENETIC INFORMATION

We plan to use advanced DNA techniques to test for changes in genes that may contribute to your child's known or suspected SMS diagnosis. The new DNA sequencing technology looks at all the human genes we know about; this is known as *genome sequencing*. We are looking to see if any of these genes have changes that cause or contribute to SMS. This research may take years. Because this new technology is much more detailed than previous DNA testing, we would like to explain what we plan to do and get your permission to do this.

1. Genome sequencing and other forms of genetic testing are research tests that can provide various types of results. For example, we can find gene variants that are known to cause or contribute to disease. We can find gene variants that are known NOT to cause or contribute to disease, meaning that they are normal variations of the genome. We can find gene variants that are novel and of uncertain clinical importance, meaning that we do not know if they cause or contribute to disease or if they are normal variations of the genome.
2. Most of the results that are important to your child's or your family's health and/or the health of you/your relatives

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NIH-2514-1 (10-84)

NIH-2514-2 (10-11)

2008

MEDICAL RECORD

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NIH 2514-1, Consent to Participate in A Clinical Research Study

NIH 2514-2, Minor Patient's Assent to Participate In A Clinical Research Study

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fall in the first category, i.e., gene variants that are known to cause or contribute to disease. If we find one or more of these gene variants in your child's DNA sample, it could be that: the gene variant has already caused health problems or may cause health problems in the future. Identifying gene changes that are not related to the disease being studied (i.e., SMS) are known as *incidental medical information*. We will find gene changes in everyone that we study, but we are only specifically looking for genes related to the disorder (SMS) being studied.

- 3. If we **find a** gene variant that has health implications, we will repeat the test for that specific gene variant in a clinical laboratory, i.e., one that is approved for supplying genetic information to patients. We will **only** give you results about specific abnormal gene variants that we think are important to your child's or your family's health, that have been confirmed in a clinical laboratory.
- 4. In the future, we may contact you by mail or phone to find out if you are interested in learning about these results or gene variants that are important to your child's health and/or family's health. We may also contact you to find out if you are interested in having additional clinical tests and evaluations at the
- 5. You will be given a choice **to learn or not to learn** the results of your/your child's genome sequencing. You may "opt out" of learning these results, and still be in the study. (The only exception to opting out is if we find a result that has **urgent** importance to your/your child's health. We plan to share this type of result with you. However, you should know that this type of result will be found rarely, and most people in this study will not have a result like this.)

In addition to knowing what we could find, we would like you to know the limitations of the genome sequencing and what we will not be able to tell you about the changes that we find.

- 1. Not all gene changes that cause disease will be detected.
- 2. Some changes that are not currently known to cause health problems will be found to cause health problems in the future. We cannot promise to be able to tell you about those changes in the future.
- 3. We will not tell you about gene changes that are not known to have health implications.
- 4. This gene sequencing cannot be substituted for diagnostic testing recommended by other physicians.

Research using genome sequencing may take years. Initially only summary results will be available. However, we may detect a genetic change that could be the associated with your child's disorder. Also, science changes quickly, and we may learn more about the human genome after we discuss the initial results. If we think this new information is important to your child's or your family's health, we may re-contact you. If we do detect an important genetic change, and you want us to tell you about it, then we will have to confirm the result first in a clinical testing laboratory. Please let us know your preference by initialing one of the statements.

_____ I would like to be contacted with information about the progress of this study and contacted about any results that might be related to my child's disorder being studied. I understand that this research will take years and that a specific cause might not be identified in my child. [For us to contact you, you have to keep us up to date on your current address and phone number.]

_____ Please do not contact me regarding the progress of this study or any specific gene change you found.

To facilitate future genetic research, your child's DNA sequence information may be deposited in NIH controlled public databases in a manner that would not allow someone to identify your child. Please understand that this information cannot be removed once it is deposited.

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NIH 2514-1 (A) (2/04)

NIH 2514-2 (A) (2/04)

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WHAT ARE THE RISKS OF THE STUDY?***Emotional and Psychological Risks***

Genetic information about you and your family may be discovered during this research project. The test results may show that you or your family have a change in your genes that causes this condition. Learning about this information may cause emotional or psychological stress.

Information about the parents may be discovered in the course of this research project. In other words, issues of adoption and paternity (fatherhood) may be discovered. Our policy is not to discuss this information with you unless it has direct medical implications for you or your family.

Unanticipated medical information. During the course of this study, it is possible (although not likely) that we will obtain unanticipated information about you or your child's health or genetic background. If this information is considered to be relevant to your/your child's health care we will provide it either to you or your referring physician.

We will not release any information about you or your family to your insurance company or employer without your permission. There may be a risk that genetic information obtained as a result of participation in research could be misused for discriminatory purposes. However, state and federal laws provide some protections against genetic discrimination. If you have any questions, please ask (Adjunct Principal Investigator) or the Principal Investigator. Researchers who will have access to genetic information about you will take measures to maintain the confidentiality of your information.

Confidentiality and availability of genome sequence data: All DNA samples will be coded. Only medical personnel involved in this study will have access to both the participant's name and DNA code. If we submit your/your child's DNA sequence to a NIH controlled public database it will be done in a manner that that would not allow someone to identify your child. You need to be aware that DNA sequence is like a fingerprint. There remains a theoretical risk of revealing your/your child's identity. We consider this risk to be extremely low, because identifying an individual based on these data would be very difficult, and because there appears to be no reason for anyone to do this.

You, your child, or a family member could be recognized if we publish a photograph, family tree, or specific details about you. We will not use your name(s) or publish identifiable photos/video clips without asking you to sign a separate consent form for photography. Unpublished photos/video clips will be used for teaching and record keeping purposes.

All of these issues should be carefully considered before joining the study. Genetic counselors are available to discuss these issues in greater detail to help you and your family think about the possible risks.

Risks from Related Evaluations/Diagnostic procedures**Blood collection**

There may be minimal discomfort involved with blood collection. A numbing cream will be applied prior to blood draws to reduce the pain associated with this procedure. There is a small chance that your child will develop a bruise or an infection at the needle site, or may feel lightheaded or faint.

Saliva collection

There may be minimal discomfort (dry mouth) associated with collecting 3-5 cc of saliva. Collection of saliva samples at home may be slightly inconvenient.

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Skin biopsy

There may be mild pain at the biopsy site for one to two days; bleeding and infection are rare. Biopsy wounds heal usually with a very small scar, but sometimes, a raised scar or a visible lump may result. The biopsy will be taken from a place on the body (such as the underside of the upper arm) that is not easily seen.

Imaging studies

There is a separate consent form for imaging studies such as an MRI that may require dye to be placed in the vein. There is a small risk of a serious allergic reaction or kidney damage. Some people may get anxious in the MRI scanner. The scan can be stopped at any time. Only x-ray imaging studies that are specifically needed for this study will be done and the risks for these procedures are very slight. For women who have started menstruation, a negative urine pregnancy test is required because pregnant subjects may not participate in this study. Please advise your doctor if you have participated in research studies at the NIH or other institutions that involved the use of radiation so that we can be sure you do not receive too much radiation.

Sedation

Sedation may be required for certain procedures. All alternative measures will be considered. There is a separate consent form for any use of sedation. If your child needs sedation, you will meet with a member of the anesthesiology team who will discuss the risks of sedation with you.

Interviews/ Questionnaires

Interviews and questionnaires addressing family relationships and illness may at times be stressful. Some of the questionnaires may cause you to think about things that are difficult, or that you have not thought about before, or that you feel uneasy about.

Neuropsychological evaluation

Your child will undergo a series of tests and activities to evaluate his/her thinking abilities, general knowledge, understanding skills, language skills, and behavior. This evaluation will be divided into a minimum of two different testing sessions, approximately 1 1/2 hours each. To make the most of the testing sessions, we will try to avoid conflicts with normal naptime, meals and snacks, etc. If at any time you feel your child cannot take part or continue in the testing, tell the psychologist and testing will be stopped or rescheduled.

ARE THERE BENEFITS TO TAKING PART IN THE STUDY?

If you agree to take part in this study, there may be no direct treatment benefit to subjects or their family from this study. We hope that the information learned from this study will benefit other families with Smith-Magenis Syndrome in the future.

We will try to answer questions with the most up to date information available. If you desire, a written summary will be provided to you by a professional involved in your clinic visit. With your written consent, documentation, along with recommendations for additional evaluations or management options, will also be provided to your local health care provider(s).

WHAT ARE MY OTHER OPTIONS?

You may choose to omit any of the recommended tests, or you may choose not to enroll at all. If you like, we will give you the name of a geneticist near your home.

WHAT IF I CHANGE MY MIND?

You may stop participating in this study at any time. You may request to have the blood and tissue samples and information collected about your child destroyed. Your decision about this study will have no effect on your eligibility for future NIH studies.

MEDICAL RECORD

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WHO ELSE WILL KNOW THAT I AM IN THIS STUDY?

Members of the SMS Research Team will have access to your/your child's information.

Clinical data and/or biological materials that we collect from your child may be shared with other researchers in the future, linked together with other information such as your child's age, gender, and ethnicity. We will share such information, but we will not give other researchers your name, address, or phone number. There will be a code to link your child's clinical (phenotypic) data and biological materials with his/her name and other personal information. The code will be stored in locked file cabinet under the control of _____, Adjunct PI.

WILL I RECEIVE PAYMENT FOR BEING IN THIS STUDY?

You will not be paid for your child's participation in this study. It is possible that research using your child's blood may enable researchers to develop medical tests or treatments that have commercial value. You will not receive any money that may result from such commercial tests or treatments.

CONFLICTS OF INTEREST

The National Institutes of Health reviews NIH staff researchers at least yearly for conflicts of interest. The following link contains details on this process: <http://ethics.od.nih.gov/forms/Protocol-Review-Guide.pdf>. You may ask your research team for additional information or a copy of the Protocol Review Guide.

This protocol includes investigators who are not NIH employees. Non-NIH investigators are expected to adhere to the principles of the Protocol Review Guide but are not required to report their personal financial holdings to the NIH.

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NIH-2514-1 (10-84)

NIH-2514-2 (10-84)

P.A.: 09-25-0099

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OTHER PERTINENT INFORMATION

1. Confidentiality. When results of an NIH research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the NIH will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, the NIH will give the insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance.

The Federal Privacy Act protects the confidentiality of your NIH medical records. However, you should know that the Act allows release of some information from your medical record without your permission, for example, if it is required by the Food and Drug Administration (FDA), members of Congress, law enforcement officials, or authorized hospital accreditation organizations.

2. Policy Regarding Research-Related Injuries. The _____ will provide short-term medical care for any injury resulting from your participation in research here. In general, no long-term medical care or financial compensation for research-related injuries will be provided by the National Institutes of Health, the Clinical Center, or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.

3. Payments. The amount of payment to research volunteers is guided by the _____ policies. In general, patients are not paid for taking part in research studies at _____. Reimbursement of travel and subsistence will be offered consistent with NIH guidelines.

4. Problems or Questions. If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the Principal Investigator _____ or Adjunct Principal Investigator, _____ Telephone: _____

Other researcher you may call is: Medical Advisory Investigator _____ Telephone _____ You may also call the (_____

5. Consent Document. Please keep a copy of this document in case you want to read it again.

COMPLETE APPROPRIATE ITEM(S) BELOW:

A. Adult Patient's Consent

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby consent to take part in this study.

Signature of Adult Patient/Legal Representative Date

Print Name

B. Parent's Permission for Minor Patient.

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby give permission for my child to take part in this study. (Attach NIH 2514-2, Minor's Assent, if applicable.)

Signature of Parent(s)/Guardian Date

Print Name

C. Child's Verbal Assent (If Applicable)

The information in the above consent was described to my child and my child agrees to participate in the study.

Signature of Parent(s)/Guardian Date Print Name

**THIS CONSENT DOCUMENT HAS BEEN APPROVED FOR USE
FROM AUGUST 8, 2013 THROUGH AUGUST 7, 2014.**

Signature of Investigator Date Signature of Witness Date

Print Name

Print Name

PATIENT IDENTIFICATION

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY (Continuation Sheet)

• Adult Patient or • Parent, for Minor Patient
NIH-2514-1 (07-09)
P.A.: 09-25-0099
File in Section 4: Protocol Consent

