The UNIVERSITY OF
CHICAGO
The Division of the Biological Sciences • University of Chicago
Medical Center

CONSENT/AUTHORIZATION BY SUBJECT FOR PARTICIPATION IN A RESEARCH PROTOCOL FOR THE BETTER UNDERSTANDING OF THEIR GENETIC CONDITION

Protocol Number: 11-0151 Name of Subject: _____

Date of Birth: _____

STUDY TITLE: Molecular Genetic Studies of Rare Orphan Genetic

Disease

Research Team: Soma Das, Ph.D.
5841 S. Maryland Ave. Room G-701 MC 0077, Chicago, IL 60637
773-834-0555

You are being asked to allow your child to participate in a research study that may help us learn more about the genetic condition for which you are being tested. This consent form describes the study, the risks and benefits of participation, as well as how your confidentiality will be maintained. Please take your time to contact us with questions and feel comfortable making a decision whether to participate or not. If you decide to participate in this study, please sign this form. Throughout this consent form, "you" will refer to you or your child, as appropriate.

WHY IS THIS STUDY BEING DONE?
You have already consented to clinical genetic testing. We are asking you to also participate in further studies. The purpose of these studies is to learn more about the genetic cause of diseases tested for in our lab, gather more information about these disorders, and experiment with new methods that may be better for testing.

WHAT IS INVOLVED IN THE STUDY?
During this study, Dr. Das and her team will collect information about you for this research. We may contact your doctor to request additional Protected Health Information (PHI), which consists of any health information related to your diagnosis (such as date of birth, medical record number, primary diagnosis, clinical features, relevant and family history, outcome). The data collected will be used to develop a database of patients being tested for genetic diseases and will be kept for the duration of the database. This study will look at how often different genetic mutations happen and clinical information related to the mutation.

When our lab is researching new genes or testing methods that are related to your diagnosis, we may include your sample, with others from similar patients in a small study before offering this new test. This data will help in directing doctors about the likelihood of a positive or negative test result in their patient. We may also use your sample to set up new methods that will improve the clinical testing in

our laboratory. Your clinical information and sample, without any identifiers, may also be shared with other researchers that are interested in this specific condition.

HOW LONG WILL I BE IN THE STUDY?
Once enrolled, you will likely remain in this study as long as your DNA sample remains in our laboratory. If you want your sample, to be removed from the study at any time, please contact us, and the sample will not be used for further studies. Existing results will remain in our database until the study ends.

WHAT ARE THE RISKS OF THE STUDY?
There are no known added risks of the research. No additional information will be obtained from you, as all of the information has already been collected as part of clinical genetic testing or evaluation by your doctor.

ARE THERE ANY BENEFITS TO TAKING PART IN THE STUDY?
If you agree to take part in this study, there may be direct medical benefit to your family. We may identify a cause for the genetic disease in your family. If a mutation is identified in your DNA, through our testing, your referring doctor will be notified and will receive a clinical report. Our study may also be helpful in finding the genetic causes of disease and will benefit doctors and patients as a group.

WHAT OTHER OPTIONS ARE THERE?
You may choose not to participate.

WHAT ARE THE COSTS?
There will be no additional costs to you or your insurance company resulting from this research study. However, you or your insurance company will be responsible for costs related to your usual medical care.

WILL I BE PAID FOR MY PARTICIPATION?
You and your child will not be paid to participate.

WHAT ABOUT PRIVACY?
Study records that identify you will be kept private. All of your personal information will be entered into a password-protected database to prevent access to non-authorized personnel. If your data is shared with other researchers, all patient identifiers will be removed. Data from this study may be used in medical journals or presentations. If results from this study or related studies are made public in a medical journal, individual patients will not be identified. If we wish to use a patient's identity in a medical journal, we will ask for your permission at that time.

As part of the study, Dr. Das and her team will report any positive results of further testing to your referring doctor and/or genetic counselor. Dr. Das may also share these results, without your name or date of birth, with other researchers.

People from the University of Chicago, including the Institutional Review Board (IRB), a committee that oversees research at the University of Chicago, may also view the records of the research. If health information is shared outside the University of Chicago, the same laws that the University of Chicago must obey may not protect your health information. Dr. Das does not have to give you any results that are not are not important to your health or your family's health at that time.

This consent form will be kept by the research team for at least six years. The study results will be kept in your child's research record and be used by the research team indefinitely. When the study ends, your personal information will be removed from all results. Any information shared with your doctor may be included in your medical record and kept forever.

WHAT ARE MY RIGHTS AS A PARTICIPANT?
Taking part in this study is optional. You may choose not to participate at any time during the study. Choosing not to participate or leaving the study will not affect your clinical testing at the University of Chicago.

If you choose to leave the study and you do not want any of your future health information to be used, you must inform Dr. Das in writing at the address on the first page. Dr. Das may still use your information that was collected before your written notice. You will be given a signed copy of this form. This consent form does not have an expiration date.

WHO DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?
If you have further questions about the study, please call 773-834-0555.

If you have any questions about your rights in this research study you may contact the IRB, which protects participants in research projects. You may reach the Committee office between 8:30 am and 5:00 pm, Monday through Friday, by calling (773) 702-6505 or by writing: IRB, University of Chicago, 5751 S. Woodlawn Ave., McGiffert Hall, Chicago, Illinois 60637.

CONSENT

I have received information about this research project and the procedures. No guarantee has been given about possible results. I will receive a signed copy of this consent form for my records.

I give my permission to participate in the above research project.

Signature of Subject: _____ Date: _____

I give my permission for my child/relative/the person I represent to participate in the above research project.

Signature of Parent/Guardian/Legally Authorized Representative: _____Date: _____