Medicine
UNIVERSITY OF TORONTO

Western

NAME:

ADDRESS:

CONSENT FOR PARTICIPATION IN A RESEARCH PROJECT
UNIVERSITY OF TORONTO (TORONTO) AND UNIVERSITY OF WESTERN ONTARIO (LONDON)

Research Consent Form (Child)

Title of research project:
Evaluating the role of 7q11.23 duplications in the pathogenesis of severe speech and language disorder

Investigators:
Lucy R. Osborne, PhD - University of Toronto
Janis Oram-Cardy, PhD S-LP (C) - University of Western Ontario

Your child is invited to participate in a study that determines whether or not the speech/language delay in your family is caused by genetic changes on chromosome 7. Some people with severe speech impairment and expressive language delay have a duplication of a region of chromosome 7. At the present time we do not know how common this genetic difference is, and this study is aimed at finding out, by searching for the chromosome 7 duplication in people with speech/language impairment.

If your child participates in this study, we will collect saliva from your child and send it for genetic testing. This new testing will examine your child's DNA from the genetic region on chromosome 7 that is known to cause speech/language impairment.

In order to decide whether or not you wish your child to be a part of this research study, you should know enough about the risks and benefits to make an informed judgment. This consent form gives you detailed information about the research study. Either Investigator will be available discuss it with you as well if you wish. This discussion should include all aspects of this research, including its purpose, the procedures that will be performed, any risks of the procedures and possible benefits. Once you understand the study, you may decide to participate, at which point you will be required to sign this consent form on behalf of your child.

Description of Procedures

How the Studies are Performed:

A saliva collection kit and instructions will be mailed out to your home. Once saliva has been collected from your child (by spitting in a small cup), you will be asked to return the sample in a pre-paid package, along with a signed copy of this consent form and the completed questionnaire about your child's speech and language abilities.

The package will be returned to Dr. Lucy Osborne at the University of Toronto, where DNA will be extracted from the saliva. The DNA sample will be tested for genetic alteration of chromosome 7.

If evidence of a chromosome 7 alteration is found, we will invite you and your child to donate 20 ml of blood (or 10 ml for small children) to be used for follow-up testing. In some cases, this research sample can be obtained while you/your child are already having blood samples drawn for clinically indicated purposes. However, you/your child may choose to donate blood just for the purpose of this research project.

The blood samples will be sent to Dr. Lucy Osborne at the University of Toronto, where they will be grown in culture and then frozen for future recovery. This process provides a renewable source of cells and avoids the need to draw more blood in the future. Follow-up testing can then be performed on these cells.

Your child's DNA will be used to check genes on chromosome 7, according to standard genetic techniques. We will not perform any additional genetic tests of other chromosomes. Dr. Osborne will give the results of her studies directly to your child's Speech Language Pathologist, general practitioner or pediatrician. In turn, your child's health professional will send you a letter that summarizes the findings or speak with you about the results during a consultation.

Should we find evidence of a chromosome 7 alteration we will also we will ask you to share information (like reports or test results) about your child's speech and language skills. We will ask for your permission to speak with your child's Speech-Language Pathologist to gather more detailed information. This will help us to see whether there are any specific characteristics shared by children who have a chromosome 7 duplication.

Risks and Inconveniences:
The risks in this study are minimal. In most cases, we will only be collecting saliva in a non-invasive manner. In a few cases we may request a follow-up blood sample. The discomforts of blood drawing can be minimized by application of EMLA cream (a numbing cream applied to the skin before the blood is drawn). Occasionally, individuals may have some black and blueness of the skin in areas where the blood was taken.

New genetic information about the nature of chromosome 7 in people with speech/language impairment may result from participation in this study. We think that ultimately this information

will help us identify the gene responsible for this disorder. In the short term, we do not believe this information will affect you/your child's daily medical care or interfere with insurability or receipt of benefits.

This knowledge could cause psychological stress to your family. In rare cases, knowing about a presence of a genetic problem might *possibly* affect your health insurance coverage in the future, although this is very unlikely.

Benefits:
This study may not provide any direct benefit to your child but will increase our understanding of the genetic basis of speech language impairment. It may also tell us important information about the role of certain genes in human brain and/or speech/language development.

Economic Considerations:
Your child will not be paid for participating in this study. You will not be billed for providing samples, shipment of samples or genetic testing. There will not be any commercialization of your child's cells but it is possible there may be commercialization of data obtained using your child's cells.

Confidentiality:
Your child's identity will be known only to the Investigators. Each saliva sample will receive a unique ID number used during DNA extraction in Dr Osborne's laboratory and at every subsequent point. When blood is sent, it will be transferred to the Tissue Culture Facility at the Hospital for Sick Children in Toronto, where a tissue culture number will be assigned. The sample will be referred to by the tissue culture number at every point during subsequent handling. Only the Investigators will have a table that correlates the patient name, ID number and tissue culture number. This table will be accessed only by them, and stored on their computers, in offices that are locked unless they are inside.

Any publication resulting from this research will maintain patient confidentiality by referring to the individuals by ID number only.

Voluntary Participation:
Your family is free to decide not to participate in this research study. If you do decide to participate, you may withdraw at any time (see below). These decisions will in no way affect your child's relationship with the University of Toronto, or your child's own hospital.

Disposition of DNA/Blood Sample:
After donating a saliva or blood sample to this research study, you may change your mind and ask that part or all of your child's sample be withdrawn from the study. You should write the Investigators a letter and request either:

a) that your child's sample be discarded so that no further work is performed on it, or
b) that your child's sample be made "anonymous" (never be traced back to your child). If your child's sample is made anonymous, it may be used for other testing but you will not receive results from this testing,
c) that your child's frozen cells be discarded.

Questions:

Please take time to read this form and make sure that you understand it. Feel free to take as much time as you need and ask any questions you may have, as some of the terms are complex. Your signature on this form indicates that you have read the above explanation of the procedure(s), and you have given voluntary consent to participate in this study and that you have received a copy of the protocol.

Authorization: I have read this form and decided that _____
(Name of subject)
will participate in the project described above. The purpose, details and possible inconveniences of the study have been explained to my satisfaction. My signature also indicates that I have received a copy of this consent form.

_____
Signature

_____ _____
Relationship (self, parent, guardian) Date

The purpose, details, and possible risks and inconveniences of the study have been explained to me.

Assent:_____
Signature of Minor

_____
Signature of Principal Investigator Telephone
Or

_____
Signature of Person Obtaining Consent Telephone

If you have further questions about this project or if you have a research-related injury, please contact your physician.