NAME:

ADDRESS:

HOSPITAL NUMBER:

CONSENT FOR PARTICIPATION IN A RESEARCH PROJECT
UNIVERSITY OF TORONTO - TORONTO

Title: Correlating Genotype and Phenotype in Williams syndrome

You/your child are invited to participate in a study that determines whether or not the genetic basis of Williams syndrome (WS) in your family differs from that found in most persons with WS. You/your child/a member of your family have already been diagnosed with WS and, in all likelihood, results from the specialized genetic testing (FISH) have confirmed this diagnosis. Most persons with WS have the same size deletion on chromosome 7, involving loss of ~25 genes. Occasionally, a person is diagnosed with WS by their doctor, but FISH does not confirm the diagnosis.

If you participate in this study, we will collect 1-2 teaspoons of blood and send it for additional genetic testing. This new testing will examine your DNA from the genetic region known to cause WS. We will see if you are missing the same or different number of genes as most persons with WS/your family member with WS.

In order to decide whether or not you wish 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. A member of our research team will discuss it with you as well. 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 will be asked if you wish to participate. If you decide to do so, you will be asked to sign this consent form.

Description of Procedures

How the Studies are Performed:
Your physician will invite you to donate an extra 20 ml of blood (adults) or 10 ml (children) to be used for this study. In most cases, this sample will be obtained while you are already having blood samples drawn according to your doctor's recommendations. However, you may choose to donate blood just for the purpose of this research project.

The blood sample will be sent to Dr. Lucy Osborne at the University of Toronto. Half the sample will be used to obtain DNA (your genetic code). The other half will be grown in culture and then frozen for future recovery. This process provides a renewable source of DNA and avoids the need to draw more blood in the future.

Your DNA will be used to check genes in the WS region, according to standard genetic techniques. We will not perform any additional genetic analyses outside the WS region located on chromosome #7. Dr. Osborne will give the results of her studies directly to your physician. In turn, your physician, will send you a letter that summarizes the findings.

Risks and Inconveniences:
The risks in this study are minimal. 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 the WS deletion region on chromosome 7 may result from participation in this study. We think that ultimately this information will help us sort out the role each gene plays in the causation of WS. In the short term, we do not believe this information will affect your daily medical care or interfere with insurability or receipt of benefits.

Benefits:
This study may not provide any direct benefit to you but will increase our understanding of the genetic basis of Williams syndrome. It may also tell us important information about the role of certain genes in human development.

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

Confidentiality:
Your identity will only be known to the investigators, your physician, and Dr. Lucy Osborne. Each sample will receive 2 unique ID numbers:

a) a temporary number used during DNA extraction in Dr Osborne's lab and

b) a permanent tissue culture number. When the sample arrives at the Tissue Culture Facility at the Hospital for Sick Children in Toronto, the facility requires information about the sample such as referring physician and patient name, as this avoids sample confusion. A tissue culture number is then assigned.

The sample will be referred to by the tissue culture number at every point during subsequent handling. Only Dr. Osborne will have a table that correlates the patient name, temporary identifier and tissue culture number. This table will accessed only be her, and stored on her computer, in an office that is locked unless she is inside.

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

Voluntary Participation:

You are 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 relationship with the physicians, nurses, The Hospital for Sick Children, the University of Toronto, or your own hospital.

Disposition of Blood Sample:

You may choose to participate in only part of this study. Specifically, you may want the DNA analysis but not the freezing of cells. This latter procedure allows us to test your sample for new developments in the genetic region causing WS without obtaining another blood sample from you. To do this, we maintain your cells indefinitely. Again, we will only do new testing on genes located on chromosome 7 in the WS region but you are free to decline participation in this portion of the study.

After donating a blood sample to this research study, you may change your mind and ask that part or all of your sample be withdrawn from the study. You should write your physician a letter and request either:

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