Medicine
UNIVERSITY OF TORONTO

FACULTY OF MEDICINE | UNIVERSITY OF CALGARY

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

Research Project: Cytogenetic analysis of the Williams-Beuren syndrome region on chromosome 7 in human sperm

Investigators: Dr. Lucy Osborne, Department of Medicine, University of Toronto

Dr. Renée Martin, Department of Medical Genetics, University of Calgary

Funding Agency: Canadian Institutes of Health Research

You are invited to participate in a study that aims to determine the effect of the common inversion at the Williams-Beuren syndrome (WBS) region, on chromosome recombination in germ cells (eggs and sperm).

We would like to gather more information in order to better understand the effect of the WBS inversion chromosome (WBSinv-1) on meiosis (the type of cell division by which germ cells are produced). It is recombination between chromosomes during this cell division that results in the deletion, or sometimes duplication, of the WBS region. We do not know whether carriers of the WBSinv-1 chromosome have more germ cells (eggs and sperm) that carry either a deletion or duplication of the WBS region.

If you decide to participate in this study, a semen sample will be sent for cytogenetic analysis to count the number of sperm cells with a rearrangement of the WBS region on chromosome 7. We will also extract DNA from the sperm to use a molecular method to estimate the frequency of deletion of the WBS region.

This consent form, a copy of which has been given to you, is only part of the process of informed consent. It should give you the basic idea of what the research project is about and what your participation will involve. If you would like more detail about something mentioned here, or information not included here, you should feel free to ask. Please take the time to read this carefully and to understand any accompanying information. Your signature on this form indicates that you have read the 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.

Purpose of Research
The purpose of this research is to investigate the mechanism by which the inversion polymorphism of the WBS region predisposes the chromosome to subsequent deletion, and to measure the frequency of chromosome rearrangements in carriers of the inversion.

A previous genetic study on WBS identified a common inversion of the region that is usually deleted in WBS (named the WBSinv-1 chromosome). This WBSinv-1 chromosome is found in up to 5% of the general population. It is found in parents of children with WBS at a much higher frequency (between 25 and 30%), suggesting that it may be a risk factor for the deletion. We aim to look at the frequency and nature of chromosome 7 rearrangements in human sperm from both carriers and non-carriers of the WBS inversion polymorphism. We plan to study four carriers, and four non-carriers of the WBS inversion, and we will analyze 20,000 individual sperm from each sample.

The results of this research will certainly benefit medical science as we have no information available on the frequency of chromosome rearrangements in the human sperm of carriers of the WBS inversion. The results will not benefit you directly but will be used to understand the process and consequences of unequal recombination in inversion carriers.

How the Studies are Performed:
We require a semen sample, produced by masturbation at home (in a sterile container which we will provide). Although arranged at your convenience, the samples must be shipped, on ice, to the research laboratory immediately. We will provide appropriate pre-paid shipping and packing materials.

The semen sample will be sent to Dr. Renée Martin at the University of Calgary where analysis of chromosome 7 will be performed. Using cytogenetic techniques to label the region of chromosome 7 with fluorescent dyes, we will study more than 20,000 individual sperm cells from a single sample, to look for rearrangements of the region. In addition, DNA will be extracted from half of the semen sample and shipped to Dr. Matthew Hurles's laboratory at the Wellcome Trust Sanger Centre, UK for the molecular analysis.

Donor confidentiality is maintained at all times. If the results of this research are published, it will not be possible to identify donors in any way. Only Dr. Osborne and Dr. Martin will have access to that information.

We also ask that you fill out a Donor Information Sheet, which will inform us of any medical or environmental history that may have had an effect on your sperm.

Risks and Inconveniences:
The risks in this study are minimal. We will only be collecting semen, which is a non-invasive procedure.

The results of this study will be analyzed as a group, not on an individual basis. Individual results from this study will not be released to participants. We will be able to provide you with a summary of the results from the entire study, once it has been completed.

New information about the frequency and mechanism of the WBS deletion on chromosome 7 may result from participation in this study. We think that ultimately this information will help us to generate more accurate risk factors for the WBS deletion in individuals who carry the WBS inversion. This information will not affect your daily medical care or interfere with insurability or receipt of benefits.

Benefits:
This study will not provide any direct benefit to you but will increase our understanding of the genetic basis of Williams-Beuren syndrome.

Economic Considerations:
You will not be paid for participating in this study. You will not be billed for shipment of samples or cytogenetic 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 only to the Principal Investigators. When the sample arrives at the University of Calgary, an identifying number will be assigned by Dr. Martin. The sample will be referred to by this number at every point during subsequent handling. Only Dr. Osborne and Dr. Martin will have a table that correlates the donor name and sample number. This table will be accessible only to them, stored on their computers in offices that are locked unless they are inside.

Any personal information provided on the DONOR INFORMATION SHEET will remain confidential and will not be disclosed under any circumstances.

Any publication resulting from this research will maintain confidentiality by referring to the individuals by identification 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. After donating a sperm sample to this research study, you may change your mind and ask that your sample be withdrawn from the study. You should write the Principal Investigator a letter and request that your sample be discarded. These decisions will in no way affect your relationship with the University of Toronto or The University of Calgary.

Your signature on the consent form indicates that you have understood to your satisfaction the information regarding your participation in the research project and agree to participate as a subject. In no way does this waive your legal rights nor release the investigators, sponsors, or involved institutions from their legal and professional responsibilities. You are free to withdraw from the study at any time without jeopardizing your health care. Your continued participation should be as informed as your initial consent, so you should feel free to ask for clarification or new information throughout your participation. If you have further questions concerning matters related to this research, please contact Dr. Lucy Osborne at 416 946 5804. If you have any questions concerning your rights as a possible participant in this research, please contact the Ethics Review Office, Faculty of Medicine, The University of Toronto, at ethics.review@utoronto.ca or 416 946-3273.

Authorization: I have read this form and decided that I, _____ will
(Name of donor)

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 of Principal Investigator	Telephone	Date
_____	_____	_____
Signature of Witness	Telephone	Date

You should keep a copy of this consent form for your records and reference.

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