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Medical Genetics, UBC
Child & Family Research Institute

## INTRODUCTION

In addition to the main part of the research study, you are being invited to participate in this optional part because you or your child has been observed to either have clinical features suggestive of an imprinting disorder or have a chromosome error that may be associated with an imbalance in gene contribution or gene regulation from the mother and father. We are interested in identifying the genetic changes associated with imprinting disorders and how the underlying cause leads to the clinical condition. This information can then be used to improve diagnosis, counselling, and eventually treatment of individuals with such disorders.

Your participation is entirely voluntary, so it is up to you to decide whether or not to take part in this part of the study. Before you decide, it is important for you to understand what the research involves. If you wish to participate, you will be invited to sign this form. If you do decide to take part in this study, you are still free to withdraw at any time and without giving any reasons for your decision. If you do not wish to participate, you do not have to provide any reason for your decision not to participate nor will you lose the benefit of any medical care to which you are entitled or are presently receiving. Please take time to read the following information carefully and to discuss it with your family, friends, and doctor before you decide. Any questions concerning participation can be addressed to Dr. Robinson at the above phone number.

## BACKGROUND

Genomic imprinting occurs when the expression of a gene depends on which parent (mother or father) each copy of the gene was inherited from. Some disorders that are associated with abnormal gene expression of imprinted genes include, among others, Russell Silver syndrome, Beckwith-Wiedemann Syndrome, transient neonatal diabetes, and hemi-hypertrophy. These disorders can arise any one of several ways; one cause can be uniparental disomy (UPD), which is the situation when both copies of a pair of chromosomes (the structures into which our genes or DNA are packaged) have come from one parent rather than the usual situation where one copy is inherited from each parent. Another way is when the gene's genetic material is altered somehow and the copy inherited from the mother behaves like the one from the father (or vice versa).

## PURPOSE

The aim of tissue banking is to understand the causes and mechanism of imprinting and imprinting disorders. Often it is difficult to anticipate future advances in science that yield new technology or new research questions. We hope to understand imprinting and its disorders by developing future research goals. We will store the de-identified DNA samples from subjects who either present clinical features or a chromosomal change that is

associated with an imprinting disorder. The study involves comparing the subject's chromosomes to his or her parents. Therefore, de-identified DNA samples from the parents, as well as the subject, for families interested in participating in this study will also be stored. Each individual participating will receive their own consent to sign. If the subject is not able to sign for themselves due to age or developmental capacity, their legally accepted guardian will sign. If the genetic parents are not able to participate, the subject may still participate.

RESULTS

Participants will not be notified of results from these future studies. In rare cases, if results are provided, the counselling regarding potential results will reside with the original health care professional who requested the study. If results are provided, they will be provided to the requesting health care provider. The health care provider will contact you to inform you that additional results are available. It is up to the health care provider to determine whether results will be provided by an in person appointment or whether a phone appointment is sufficient.

RISKS

In the rare cases where results are provided, there is the potential for identifying an underlying genetic change associated with clinical features. There may be psychological harm associated with identification of a genetic change that cannot be reversed, treated or cured. An additional risk inherent with genetic studies involving parents and a child is the risk of identifying non-paternity. In situations where non-paternity is identified it may cause psychological distress for the family. For risks associated with genetic testing in general please visit www.humgene.org.

WITHDRAWING CONSENT

Your participation in this optional part of the research is entirely voluntary. If you decide to enter the study and later decide to withdraw at any time in the future, there will be no penalty or loss of benefits to which you are otherwise entitled, and your future medical care will not be affected. Furthermore, your eligibility to participate in the main part of the study will not be affected. If you wish for your stored blood samples/data to be destroyed upon your withdrawal, you may contact the principal investigator of the study, at 604.875.3229. The investigator may decide to discontinue the study at any time, or withdraw you from the study at any time. If you choose to enter the study and then decide to withdraw at a later time, all data collected about you during your enrolment in the study and up to the time of withdrawal will be retained for analysis. By law, this data cannot be destroyed.

CONFIDENTIALITY

In Canada genetic information as a form of personal information is protected legally by privacy and discrimination Acts. Your confidentiality will be respected. No information that discloses your identity will be released or published without your specific consent to the

disclosure. However, research records and medical records identifying you may be inspected in the presence of the Investigator or his or her designate, Health Canada, and the UBC Research Ethics Boards for the purpose of monitoring the research. However, no records which identify you by name or initials will be allowed to leave the Investigators' offices.

Dr. Robinson is the custodian of the list of participant names and the linking code. Dr. Robinson may provide access to the list of names and linking code to the research coordinator and lab manager. Both the research coordinator and lab manager have signed confidentiality agreements. The samples will be de-identified and coded upon receipt. The list of names is in a password protected electronic file. The electronic file is kept on the lab server only accessible with an authorized computer login and password.

No further consent will be sought from you for these future related genetic studies. Ethics approval will be sought for any additional research goals. Your de-identified DNA will be stored in Dr. Robinson's laboratory at the Child & Family Research Institute (CFRI). The de-identified DNA sample labelled with the code will be stored until it is used entirely or until such DNA is withdrawn. The laboratory is in a secure building accessible only by photo key card.

Should your de-identified sample be requested by an outside investigator Dr. Robinson will obtain the proper ethics approval prior to donating the de-identified sample. Dr. Robinson will ensure that the outside investigator has the proper ethics approval from their institution. Samples will only be donated if the outside investigator's research goals involve genomic imprinting & its disorders. At no point will any identifiable data associated with the de-identified sample be sent to outside investigators. Your de-identified DNA sample will not be sold and will not be used for commercial purposes.

Signing this consent form in no way limits your legal rights against the sponsor, investigators, or anyone else. Your rights to privacy are also protected by the *Freedom of Information and Protection of Privacy Act of British Columbia.* This Act outlines rules for the collection, protection, and retention of your personal information by public bodies, such as the University of British Columbia and its affiliated teaching hospitals. Further details about this Act are available upon request.

CONSENT FOR A MINOR

If you are consenting for your child, who is under 19 years of age, Dr. Robinson's lab will attempt to contact your child when he or she reaches 19 years of age. The purpose for contacting your child in the future is to obtain his or her consent for the study. If you change your contact information without updating us, please have your child re-contact the lab when he or she reaches 19 years of age. We will provide your adult child with their own consent form. If your adult child wishes to withdraw from the study the above information in the "withdrawing consent" section will apply. Dr. Robinson can be contacted at 604.875.3229.

CONSENT TO PARTICIPATE

If you have any concerns about your rights as a research subject and/or your experiences while participating in this study, contact the Research Subject Information Line in the University of British Columbia Office of Research Services at 604-822-8598.

- I have read and understood the subject information and consent form.
- I have had sufficient time to consider the information provided and to ask for advice if necessary.
- I have had the opportunity to ask questions and have had satisfactory responses to my questions.
- I understand that all of the information collected will be kept confidential and that the result will only be used for scientific objectives.
- I understand that my participation in this study is voluntary and that I am completely free to refuse to participate or to withdraw from this study at any time without effecting my participation in the main study and without changing in any way the quality of care that I receive.
- I understand that I am not waiving any of my legal rights as a result of signing this consent form.
- I understand that there is no guarantee that this study will provide any benefits to me
- I have read this form and I freely consent to participate in this study.
- I have been told that I will receive a dated and signed copy of this form.

Bank de-identified DNA for genetic studies related to genomic imprinting & its disorders:

☐ **all** future studies
OR

☐ only involving the use of **new technology**.
OR

☐ only performed by **UBC investigators**.

| Printed name of subject | Signature | Date |
|---|---|---|
| Printed name of subject's Legally Acceptable Representative (if applicable) | Signature | Date |
| Printed name of Principal Investigator/ designated representative | Signature | Date |