Parental Informed Consent Form
Prenatally detected Chromosome Mosaicism

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Background: Prenatal diagnosis of your pregnancy has recently revealed the presence of a chromosomal abnormality in a portion of cells obtained by amniocentesis or chorionic villus sampling. The presence of a chromosome abnormality in some but not all cells is called chromosomal mosaicism. The chromosome abnormality may have been confined only to placental tissue or may be present in the fetus as well.

Aim: The purpose of this research proposal is to improve our prediction of outcome in such pregnancies by comparing cytogenetic (studies of the whole chromosomes) and molecular (studies of genetic markers along the chromosome) analysis with the course and outcome of the pregnancy. The cytogenetic studies involve evaluating the distribution of the abnormal cell line and its origin using placental samples from any pregnancies that have been identified to show chromosomal mosaicism. Fetal samples will also be examined in the case of pregnancy termination. The molecular analysis compares the placental and/or fetal chromosomes to the parents in order to understand how the chromosome abnormality arose. Therefore, blood must also be taken from the parents. Clinical information including weight, sex and gestational age of the baby will be obtained from medical records.

Procedure: Four 4cc (approximately 1.5 teaspoonfuls) tubes of blood will be drawn from **each** parent for these genetic studies. In addition, a sample of placental tissue (and fetal tissue if relevant) from the pregnancy will be used for genetic studies of the origin of the abnormal chromosome. Participation includes signing a release of information form for clinical records.

RESULTS: Participants may be provided with results from these studies. We do not have an estimated turn around time for the results as this study is unfunded and completed on a volunteer basis by lab personnel. When results are provided, the counselling regarding results will reside with the original health care professional who requested the study. Results will be provided to the requesting health care provider. The health care provider will contact you to inform you that the 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: There may be some discomfort associated with the placement of the needle for blood withdrawal and occasionally bruising, swelling, feeling faint or dizzy and/or the rare chance an infection may result.

In addition to the risks of physical harms outlined in this consent form, there are also possible non-physical risks associated with taking part in this study. For example, disclosure of genetic or tissue marker research data could result in discrimination by employers or insurance providers toward you or your biological (blood) relatives. However in Canada genetic information as a form of personal information is protected legally by Privacy and Discrimination Acts. The chance that research data would be released is estimated to be very small.

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.

Benefits: Your participation in this study will help to further our understanding of chromosome mosaicism: how it arises and prediction of pregnancy outcome.

Compensation: There will be no compensation for your participation in this research study.

Withdrawing Consent: Your participation in this 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. If you wish for your stored blood, placenta 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. However, research records and medical records identifying you may be inspected in the presence of the Investigator or his or her designated representatives, Health Canada, and the UBC and CW Research Ethics Boards for the purpose of monitoring the research. No information or records that disclose your identity will be published without your consent, nor will any information or records that disclose your identity be removed or released without your consent unless required by law.

You will be assigned a unique study number as a subject in this study. Only this number will be used on any research-related information collected about you during the course of this study, so that your identity [i.e. your name or any other information that could identify you] as a subject in this study will be kept confidential. Information that contains your identity will remain only with the Principal Investigator and/or designate. The list that matches your name to the unique identifier that is used on your research-related information will not be removed or released without your consent unless required by law. The list the matches your name with the unique identifier 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.

Your rights to privacy are legally protected by federal and provincial laws that require safeguards to insure that your privacy is respected and also give you the right of access to the information about you that has been provided to the sponsor and, if need be, an opportunity to correct any errors in this information. Further details about these laws are available on request to your study doctor.

Your de-identified DNA will be stored in Dr. Robinson's laboratory at the Child & Family Research Institute (CFRI). The de-identified DNA sample labeled with the code will be stored until our current research goals are completed or 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 in collaboration with Dr. Robinson, she will obtain the proper ethics approval prior to sharing the de-identified sample. Should

Dr. Robinson donate a portion of your sample to an outside investigator she 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 are in keeping with the types of research that you wish your sample to be used for, as indicated on the tissue banking consent form. 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.

Any study related data/samples, sent outside of Canadian borders may increase the risk of disclosure of information because the laws in those countries *[for example, the Patriot Act in the United States]* dealing with protection of information may not be as strict as in Canada. However, all study related data/samples, that might transferred outside of Canada will be coded (this means it will not contain your name or personal identifying information) before leaving the study site. By signing this consent form, you are consenting to the possible transfer of your information and/or samples, to organizations located outside of Canada.

We invite you to bank your de-identified DNA sample received from this study for use in future related studies. A separate optional Tissue Banking Consent Form will be provided to you.

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.

Your signature on this form signifies that you consent to participate in this study and bank your de-identified DNA sample for the duration of the study. After reading the above information, you have been given the opportunity to discuss pertinent aspects of this research study and to ask questions and that you have also received a signed and dated copy of this consent form for your own records.

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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 by e-mail at RSIL@ors.ubc.ca or by phone at 604-822-8598 (Toll Free number 1-877-822-8598).

- *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 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 (if applicable).*
- *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.*

Please check below

□ I agree that a member of Dr. Robinson's research team may contact me in the future for follow-up or further research related to this study.
□ I agree that a member of Dr. Robinson's research team may contact me in the future to ask if I am interested in participating in other research studies not described in this form.

Print name of subject	Signature	Date

Print name of person obtaining consent	Signature	Date