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INVITATION:
Your have been invited to participate in this research study because you or a family member have been diagnosed with a genetic condition or an X chromosome abnormality. Your participation is voluntary. You have the right to refuse to participate in this study. If you decide to participate, you may still choose to leave the study at any time without any negative consequences to the medical care, education, or other services you are entitled or are presently receiving. Before you decide, it is important for you to understand what the research involves. This assent form will tell you about the study, why the research is being done, what will happen to you during the study and the possible benefits, risks and discomforts. If you wish to participate in this study, you will be asked to sign this form. Please take time to read the following information carefully and to discuss it with your family, friends, and doctor before you decide.

BACKGROUND
Females have 2 X chromosomes and Males have 1 X chromosome. X-chromosome inactivation (XCI) is a normal to compensate for the extra X chromosome in females. XCI involves the inactivation of most genes (not all) on one of the two X-chromosomes. In an individual the process is generally random (one X is activated in 50% of the cells and the other X is activated in the other 50% of the cells). Mosaicism is the term used to describe two different populations of cells. This non-random XCI is termed XCI Skewing. Mosaicism within an individual for an activated or inactivated X chromosome is common because it is generally a random process. However sometimes one tissue type may show the normal random (non-skewed) XCI but a different tissue type may show non-random (skewed) XCI. Non-random XCI can be linked to health problems. For decades, XCI has been tested using a method associated with the Androgen Receptor (*AR*) gene located on the X chromosome. Most chromosomes have a long arm "q" and a short arm "p". The AR gene is located on q arm and the test does not always work . Our goal is to develop new tests along both chromosome arms that can be used in other labs.

PURPOSE:
The goal is to develop several novel tests along both the X p and q chromosome arms, such that not only can non-random XCI be assessed in multiple tissue types but also the direction of skewing at different regions along the X-chromosome arms.

WHAT THE STUDY INVOLVES:
Participation involves release of medical/health records related to the genetic diagnosis or X chromosome abnormalities in your or a family member (lab/pathology report on any tissues, ultrasound reports, Medical Genetics/Specialty consult notes). The medical records allow us to verify the results we derive from the studies.

Participation also involves donation of any samples sent to pathology for analysis and donation of four tubes (approximately 2 teaspoons) of blood from each family member.

The amount of time needed is approximately 20 minutes. Each participant is to receive his/her own consent form.

POSSIBLE HARMS & DISCOMFORTS:
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.

RESULTS:
Results will be provided to your doctor or genetic counsellor when requested to confirm a diagnosis or for counselling. When results are provided, the counselling will be provided by your doctor who requested the study.

BENEFITS
Participation in this study may help to increase our understanding of the X chromosome and associated conditions.

REMUNERATION
There will be no payment for your participation in this research study.

WITHDRAWING CONSENT: Your participation in this research is entirely voluntary. Nobody will be angry if you decide to enter the study and later decide to withdraw at any time in the future. If you wish for your stored samples/data to be destroyed when you leave the study, you may contact us at 604.875.3015.

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. 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. We will be collecting personal identifiers such as year and month of birth because many genetic factors are associated with age and prevent sample mis-handling.

You will be assigned a unique study number as a subject in this study so that your identity as a subject in this study will be kept confidential. The list that matches your name to the unique study number will not be removed or released without your consent unless required by law. The list that matches your name to the unique study number 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 de-identified samples and DNA will be stored in Dr. Robinson's laboratory at the Child & Family Research Institute (CFRI). The de-identified samples labelled with the unique study code will be stored until it is used entirely or until it is withdrawn. The laboratory is in a secure building accessible only by photo key card.

WHO DO I CONTACT IF I HAVE ANY QUESTIONS OR CONCERNS ABOUT PARTICIPATION IN THIS STUDY?

If you have any questions about your participation in this study contact the Robinson Lab at 604.875.3015.

WHO DO I CONTACT IF I HAVE ANY QUESTIONS OR CONCERNS ABOUT MY RIGHTS AS A SUBJECT?

If you have any concerns or complaints 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: 1-877-822-8598).

CONSENT TO PARTICIPATE:

· 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 authorize access to my health record and samples as described in this consent form.*
- *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.

□ A member of Dr. Robinson's research team may contact me in the future for follow-up related to this study.

□ A member of Dr. Robinson's research team may contact me in the future regarding potential future research and for general information.

Print name of subject	Signature	Date