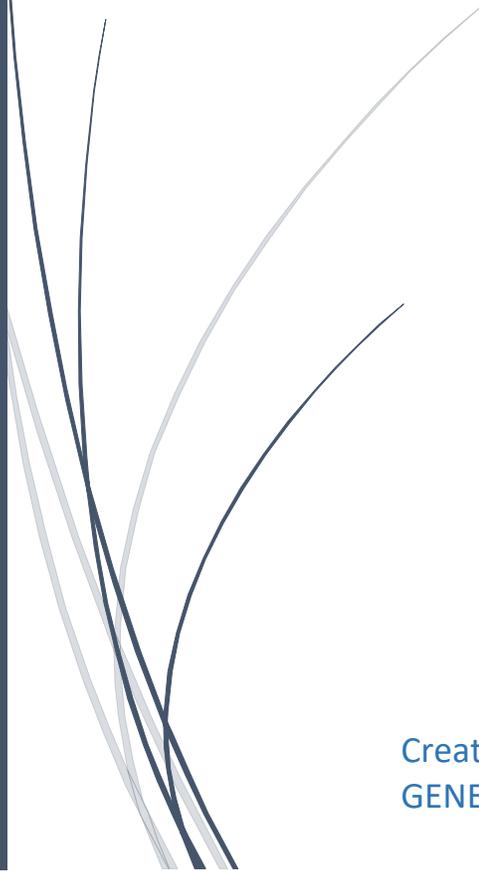




v.2 – August 21, 2019

# Genetic Counselling Guide

*For: 'Precision Diagnosis for Indigenous Families  
with Genetic Conditions' study  
(Activity 2 of 'Silent Genomes' Project)*



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GENETIC COUNSELLORS, SILENT GENOMES PROJECT

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## INTRODUCTION

The aim of this document is to support genetic counsellors and other healthcare professionals working on the research study ‘*Precision Diagnosis for Indigenous Families with Genetic Conditions*’ (herein referred to as the ‘*Precision Diagnosis*’ study). This study is Activity 2 of a larger project, ‘*Silent Genomes: Reducing Healthcare Disparities and Improving Diagnostic Success for Indigenous Children with Genetic Disease*’ (<https://bcchr.ca/silent-genomes-project>).

This guide provides an overview of the training, guiding principles, attitudes, and responsibilities required of the study team to foster cultural safety for the Indigenous families taking part in this study. It also includes resource materials and a detailed plan for the study enrollment session, and thus serves as a practical guide for study genetic counsellors.

This guide was developed by Sarah McIntosh and Karen Jacob, *Precision Diagnosis* study genetic counsellors in British Columbia, with valuable contributions from Laurie Montour and Brittany Morgan, community engagement coordinators with the *Silent Genomes* project. Special thanks to Leah Ballantyne and Kennedy Borle for their review and feedback, and to Lawrence Gillman for his help with formatting.

*We encourage the use of Parts 1 and 2 of this document for educational purposes (including presentations), as long as the source and authors are acknowledged. Editing or publication of this document may only be done with the written permission of the lead authors. Part 3 is only for internal use of the Silent Genomes study team.* © Sarah McIntosh and Karen Jacob, 2019

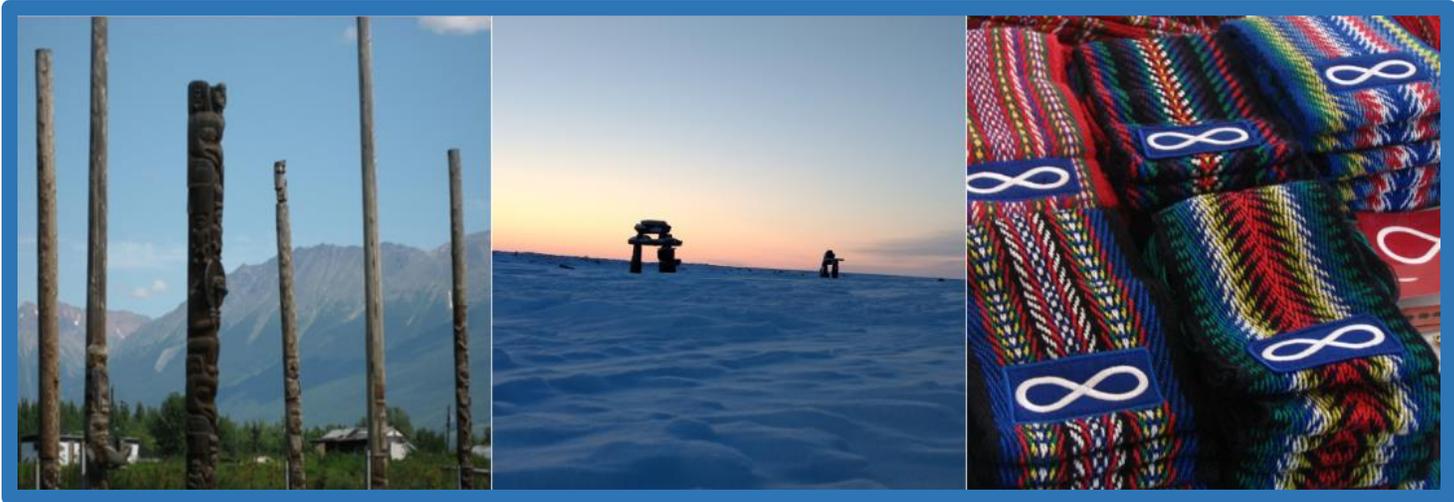
**Our partners for the overall *Silent Genomes* Project are:**



**The *Precision Diagnosis* study is funded by:**



# PART 1: INDIGENOUS CULTURAL SAFETY



## PART 1: INDIGENOUS CULTURAL SAFETY

It is well documented that Indigenous Peoples\* in Canada experience a disproportionate burden of health issues due to colonization, which has contributed to intergenerational trauma, systemic racism, oppression, and government policies that continue to exist today to perpetuate these inequities. In the current era of genomic medicine, there is significant potential for further widening of the health gap, due to Indigenous Peoples having barriers to accessing genomic technologies and precision diagnosis.

\*Throughout this guide, *Indigenous Peoples* includes First Nations (status and non-status), Inuit, and Métis people.

### ***Yes, but why is this relevant to our research study?...***

The overarching goal of the *Precision Diagnosis* study, and the larger *Silent Genomes* Project in which it is embedded, is to **promote equity in genomic diagnosis** for Indigenous Peoples in Canada, specifically by increasing access to genomic medicine. By offering whole genome sequencing technology and the possibility of precision diagnosis and improved healthcare to 200 Indigenous (First Nations, Inuit, and Métis) families across Canada with undiagnosed monogenic diseases, the *Precision Diagnosis* study is one step towards bridging the ‘genomics divide’. Given the social, political and historical contexts of Indigenous health and healthcare delivery, it is imperative that Genetic Counsellors and other healthcare providers engaging with the families participating in this study are committed to the delivery of **culturally safe care**. Cultural safety is the backbone of the *Precision Diagnosis* study and, without it, the study and its goal of reducing health disparities will not be achievable.

## **I. What is Indigenous Cultural Safety?**

**Cultural Safety** is a broad concept, first developed in New Zealand in the 1980s in response to Indigenous Māori Peoples’ negative experiences in the healthcare system. It is an approach that considers how social and historical contexts, as well as structural and interpersonal power imbalances, shape health and health care experiences. It focuses on the **systemic issues** driving health inequality, and requires healthcare providers to consider as the subtle and overt forms of racism, discrimination and prejudice and engage in **self-reflection** about how their own attitudes, beliefs, assumptions and values influence their practice.<sup>1</sup>

Cultural safety takes us beyond cultural awareness, which simply acknowledges differences between cultures, and cultural sensitivity, which recognizes the importance of respecting differences. It also surpasses cultural competence, which reduces culture into a set of ‘skills’ and knowledge for the practitioner to learn, and cultural humility, which focuses on a practitioner being a humble and open learner. All of these other approaches are incorporated into cultural safety, but none are sufficient in themselves to create cultural safety (see Table 1).<sup>1,2</sup>

Learning and becoming aware of cultural safety does not necessarily make an individual or organization ‘culturally safe’. For healthcare practitioners, cultural safety requires an ongoing commitment to **lifelong learning**. It is something we will continue to work at – a process that never stops.

It is worth noting that the continued self-reflection and learning, and the commitment to social justice and fairness required for becoming a culturally safe practitioner, are also key aspects of the *Code of Ethics for Canadian Genetic Counsellors*.<sup>3</sup>



**Table 1. How cultural safety differs from other concepts.** Adapted from Ward, C et al (2016)<sup>2</sup>

Concept	Description
<b>Cultural Awareness</b>	An attitude that includes awareness of the differences between cultures.
<b>Cultural Sensitivity</b>	An attitude that recognizes the differences between cultures and that these differences are important to acknowledge in health care.
<b>Cultural Competency</b>	An approach that focuses on practitioners attaining skills, knowledge, and attitudes to work in more effective and respectful ways with people of different cultures.
<b>Cultural Humility</b>	An approach to health care based on humble acknowledgement of oneself as a learner when it comes to understanding a person’s experience. A life-long process of learning and being self-reflective.
<b>Cultural Safety</b>	An approach that incorporates all concepts above, but also considers how social and historical contexts, as well as structural and interpersonal power imbalances, shape health and health care experiences.  Practitioners are self-reflective/self-aware with regard to their position of power and the impact of this role in relation to patients.  “Safety” is defined by those who receive the service, not by those who provide it.

## II. How Can I Facilitate Cultural Safety for Indigenous Clients/Patients?

Genetic Counsellors, as patient-centred practitioners and health advocates skilled at building respectful non-directive relationships with culturally diverse clients, are uniquely poised to facilitate Cultural Safety for Indigenous clients. However, extra skills and learning are necessary to understand Canada's damaging history with Indigenous Peoples, how this shapes health outcomes and healthcare experiences to the present day, and to commit to a path of reconciliation and health partnership.

### What can I do, as a Genetic Counsellor?

#### 1. Complete an Indigenous Cultural Safety (ICS) in Healthcare course

offered through your Institution, or by an Indigenous organization.

- ICS courses bring awareness to the social and historical contexts and power imbalances shaping Indigenous health outcomes and healthcare experiences, and promote a practice of self-reflection, humility, and lifelong learning.
- This will be your introduction to cultural safety...the groundwork that you can build further on.

Taking an ICS Course is required!

All genetic counsellors or research coordinators across Canada who will be engaging with families in the *Precision Diagnosis* study are required to take an ICS in Healthcare course.

- As soon as you are hired for the *Precision Diagnosis* study, please **contact Sarah McIntosh** (*Precision Diagnosis* genetic counsellor in Victoria, BC) at [sarahmc@uvic.ca](mailto:sarahmc@uvic.ca) to organize your ICS training.
- If an adequate ICS course is not available at your Institution, Sarah will arrange for your enrollment in the **San'yas Core ICS Health** online course. <http://www.sanyas.ca/>

#### 2. Take responsibility for your own learning.<sup>2</sup>

Read, reflect and ask questions. Commit to *lifelong learning*.<sup>2</sup>

Be prepared to be uncomfortable.

Understanding our history of colonialism, the impact of systemic racism and damaging government policies, and the history of unethical research in Indigenous populations is an ongoing and difficult task.<sup>2</sup>

- Learn about the Indigenous communities that your healthcare institution serves. *What are the names of those communities? Population size? What languages are spoken? What proportion of people are residing on- and off-reserve?*

Readings & Resources

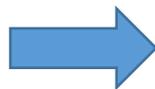
See **section IV (p.10-16)** for a list of readings and other resources to facilitate learning and cultural safety. Please note that completion of some of these resources is **required**, while others are suggested to enhance your practice.

- Learn whose traditional Indigenous territory your home and healthcare institution are located on – *what is the name of the First Nation or community whose lands you now occupy? How did your home, institution, city end up being on this land?*
- Learn about the residential Schools and ‘Indian hospitals’ that existed in your province or territory. *What was the name of the residential School and Indian hospital located closest to you? What dates were they in operation? Are you familiar with some of the stories of those institutions?*
- Learn about the health statistics in your Province, region or Institution – *how do they differ between Indigenous and non-Indigenous peoples?*
- Learn about the continuing education opportunities available to you. *Are there any Indigenous organizations or cultural safety educators available in your area to provide an educational session in your workplace?* Participate in webinars, presentations, podcasts, and other events focused on Indigenous health and cultural safety (see ‘Readings and Resources’ in Section IV for suggestions).

**3. Identify resources available to Indigenous clients/patients in your area.**

- Is there an Indigenous patient liaison or patient navigator at your healthcare institution? If so, introduce yourself and find out what his/her role is and how they may be able to help your clients/patients.
- Are there culturally safe spaces at your institution for Indigenous clients?
- Find out what healthcare support & other support services are available in the Indigenous communities near you.

**4. Get comfortable acknowledging traditional territories.**



**Traditional Territory Acknowledgement**

See if your academic institution is listed in the Canadian Association of University Teachers (CAUT) ‘**Guide to Acknowledging First Peoples and Traditional Territory**’<sup>4</sup>

Example of Territory acknowledgement for the University of Victoria:

*‘We acknowledge with respect the Lkwungen-speaking peoples on whose traditional territory the university stands and the Songhees, Esquimalt and WSÁNEĆ peoples whose historical relationships with the land continue to this day.’*

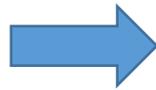
- Find out what the protocol is for acknowledging traditional Indigenous territory(ies) during presentations or events.
- On one hand, it may be preferred that every speaker acknowledges the territory on which they stand/work at the beginning of each presentation or event. On the other hand, it may be protocol that only an Elder or other leader acknowledges the territory.

**5. Take time for self-reflection.**<sup>2,5</sup>

- Get to know your own assumptions and biases. Question everything you have ‘learned’ about Indigenous people and take steps to actively disrupt the stereotypes.<sup>2</sup>

- After each session with a research participant, reflect on your practice by asking questions such as:

- Was I inclusive?
- Was I respectful?
- What more can I learn?
- What can I do better next time?



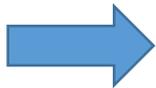
### You Will Get Better at This!

You won't be perfect. We all make mistakes. We all have a lot to learn. Being 'culturally safe' is not a place one arrives at, but rather a lifelong journey.

## 6. Reach out for help.

- Don't be afraid to ask for help if you are unsure about how to go about something, or if you are struggling with the weight of your learning and responsibilities. We all get overwhelmed at times.
- Sarah McIntosh, *Precision Diagnosis* genetic counsellor in Victoria, is available as a mentor to talk through issues without judgement. You may also find local support from other colleagues or the Indigenous Patient Liaison(s) at your institution.
- Identify who you can reach out to. It may be an Indigenous co-worker or partner, but please keep in mind to not make them feel 'tokenized'. It may also be a non-Indigenous co-worker who has years of experience being a strong ally and advocate for Indigenous Peoples and communities.
- Indigeneity is very diverse and certain advice and protocols may differ between Indigenous Peoples, communities, and regions. Do not assume that advice or approval from one Indigenous person represents all Indigenous voices.

## 7. Put cultural safety into action! <sup>2,5</sup>



- Systems don't change. Individual people change and then change the system!
- Even small changes in how you do things can add up to a big change over time.
- Your words and actions are observed by others and set the tone.

### Translate Learning into Action – 'It starts with me' <sup>3</sup>

There are many small, simple opportunities for action at every turn - you just have to look for them. Some ideas...

*Reflect upon what you can personally do, and what your workplace can do to respond to the Truth & Reconciliation Commission's 'Calls to Action'.*

*Speak out against racism & support others who do.*

*Advocate for culturally safe approaches and protocols and explain why they work for you and your clients.*

*Challenge barriers to cultural safety.*

### III. How Will We Know if Indigenous Cultural Safety Has Been Achieved?

Cultural safety is an outcome defined and experienced by those *receiving the service*. Therefore, whether cultural safety has been achieved within the *Precision Diagnosis* study can only be voiced by the Indigenous research participants themselves. If participants feel safe and empowered as a result of engaging in trusting and respectful partnerships with our research team, then cultural safety will have been achieved.<sup>1,2</sup>

#### REFERENCES – Part 1: Indigenous Cultural Safety

1. Bourassa C, McElhaney J and Oleson E. **Cultural Safety**. Webpublish Queen's University, October 2016. Weblink: [https://www.queensu.ca/sps/sites/webpublish.queensu.ca.spswww/files/files/Events/Conferences/RCAP/Papers/Bourassa\\_RCAP\\_conceptsOct2016.pdf](https://www.queensu.ca/sps/sites/webpublish.queensu.ca.spswww/files/files/Events/Conferences/RCAP/Papers/Bourassa_RCAP_conceptsOct2016.pdf). Accessed 11 October 2018.
2. Ward C, Branch C and Fridkin A. **What is Indigenous cultural safety - and why should I care about it?** *Visions*. 2016, 206: 11(4):29-32. Weblink: <http://www.heretohelp.bc.ca/visions/indigenous-people-vol11/what-is-indigenous-cultural-safety-and-why-should-i-care-about-it>. Accessed 11 October 2018.
3. Canadian Association of Genetic Counsellors. **Code of ethics for Canadian genetic counsellors**. 2006. Weblink: <https://www.cagc-accg.ca/doc/code%20of%20ethics%20e-070628.pdf>. Accessed 29 October 2018.
4. Canadian Association of University Teachers (CAUT). **Guide to Acknowledging First Peoples and traditional territory**. Weblink: <https://www.caut.ca/content/guide-acknowledging-first-peoples-traditional-territory>. Accessed 29 October 2018.
5. First Nations Health Authority. **#itstartswithme. Cultural safety and humility: Key drivers and ideas for change**. Weblink: <http://www.fnha.ca/Documents/FNHA-Cultural-Safety-and-Humility-Key-Drivers-and-Ideas-for-Change.pdf>. Accessed 11 October 2018.

#### IV. READINGS AND RESOURCES

Table 2. A list of resources that are required ( ✓ ) or suggested for Genetic Counsellors working on the *Precision Diagnosis* study.

Indigenous Cultural Safety	Description	Required?
<p><b>Indigenous Cultural Safety (ICS) in Health course</b>            For example, San'yas Core ICS Health online course: <a href="http://www.sanyas.ca/">http://www.sanyas.ca/</a></p>	<p>As soon as you are hired for the <i>Precision Diagnosis</i> study, please <b>contact Sarah McIntosh</b> at <a href="mailto:sarahmc@uvic.ca">sarahmc@uvic.ca</a> to organize your ICS training.</p> <p>Typically ICS courses are online, self-paced courses that include a total of ~8-10 hours training.            If an adequate ICS course is not available at your Institution, Sarah will arrange for your enrollment in the <b>San'yas Core ICS Health</b> online course.</p>	<p style="text-align: center;">✓</p>
<p><b>Truth and Reconciliation Commission (TRC) of Canada:</b></p> <p><i>Calls to Action</i> (2015)  <a href="http://www.trc.ca/websites/trcinstitution/File/2015/Findings/Calls_to_Action_English2.pdf">http://www.trc.ca/websites/trcinstitution/File/2015/Findings/Calls_to_Action_English2.pdf</a></p> <p><i>Honoring the Truth, Reconciling for the Future: Summary of the Final Report of the TRC</i> (2015)  <a href="http://www.trc.ca/websites/trcinstitution/File/2015/Findings/Exec_Summary_2015_05_31_web_o.pdf">http://www.trc.ca/websites/trcinstitution/File/2015/Findings/Exec_Summary_2015_05_31_web_o.pdf</a></p> <p>More resources available at: <a href="http://www.trc.ca/websites/trcinstitution/index.php?p=3">http://www.trc.ca/websites/trcinstitution/index.php?p=3</a></p>	<p><i>Calls to Action</i>: specific calls for action to redress the legacy of residential schools and advance reconciliation in Canada  <b>*pay special attention to the HEALTH Calls (#18-24)*</b></p> <p><i>Summary of Final Report</i>: review of the history and legacy of residential schools, and a summary of the findings of the TRC</p>	<p style="text-align: center;">✓</p>

<p>Ward C, Branch C and Fridkin A. <b>What is cultural safety- and why should I care about it?</b> <i>Visions</i>. 2016, 11(4):29-32.  <a href="http://www.heretohelp.bc.ca/sites/default/files/visions-indigenous-people-vol11.pdf">http://www.heretohelp.bc.ca/sites/default/files/visions-indigenous-people-vol11.pdf</a></p>	<p>Explains what cultural safety is, and why it is important.</p>	
<p><b>Podcasts:</b></p> <p><b>CBC The Current (21 June 2016).</b> <i>Meet Dr. Nadine Caron, Canada’s First Female First Nations Surgeon.</i> <a href="https://www.cbc.ca/radio/thecurrent/the-current-for-june-21-2016-1.3644974/meet-dr-nadine-caron-canada-s-first-female-first-nations-surgeon-1.3645029">https://www.cbc.ca/radio/thecurrent/the-current-for-june-21-2016-1.3644974/meet-dr-nadine-caron-canada-s-first-female-first-nations-surgeon-1.3645029</a></p> <p><b>CBC The Current (10 March 2017).</b> Dr. Michael Kirlew – ‘Our Complacency Will be Paid for in Full with Children’s Lives’  <a href="https://www.cbc.ca/radio/thecurrent/the-current-for-march-10-2017-1.4017653/our-complacency-will-be-paid-for-in-full-with-children-s-lives-warns-indigenous-health-care-advocate-1.4017780">https://www.cbc.ca/radio/thecurrent/the-current-for-march-10-2017-1.4017653/our-complacency-will-be-paid-for-in-full-with-children-s-lives-warns-indigenous-health-care-advocate-1.4017780</a></p> <p><b>CBC The Current (04 April 2018).</b> Dr. Shannon McDonald - <i>You don't belong here': Integrating Indigenous 'cultural humility' into health care</i>  <a href="https://www.cbc.ca/radio/thecurrent/features/facing-race/you-don-t-belong-here-integrating-indigenous-cultural-humility-into-health-care-1.4520698">https://www.cbc.ca/radio/thecurrent/features/facing-race/you-don-t-belong-here-integrating-indigenous-cultural-humility-into-health-care-1.4520698</a></p> <p><b>CBC The Current (29 January 2013).</b> <i>State of care: Canada’s segregated health care.</i> Documentary, Stephanie Law.  <a href="https://www.cbc.ca/radio/thecurrent/state-of-care-documentary-canada-s-segregated-health-care-1.1470423">https://www.cbc.ca/radio/thecurrent/state-of-care-documentary-canada-s-segregated-health-care-1.1470423</a></p> <p><b>CBC The Sunday Edition (05 August 2016).</b> <i>Canada: The story of a separate and unequal healthcare system</i>  <a href="https://www.cbc.ca/radio/thesundayedition/revolver-at-50-canada-s-history-of-segregated-healthcare-frog-march-firings-penny-lang-1.3707673/the-story-of-a-separate-and-unequal-canadian-health-care-system-1.3707678">https://www.cbc.ca/radio/thesundayedition/revolver-at-50-canada-s-history-of-segregated-healthcare-frog-march-firings-penny-lang-1.3707673/the-story-of-a-separate-and-unequal-canadian-health-care-system-1.3707678</a></p>	<p>Radio interviews of physicians, advocates, and journalists sharing the truths, challenges, and future hopes for Indigenous health and reconciliation.</p>	

<p><b>Webinars:</b></p> <p><b>First Nations Health Authority- Cultural Safety and Cultural Humility Webinar Series:</b>  <a href="http://www.fnha.ca/wellness/cultural-humility/webinars">http://www.fnha.ca/wellness/cultural-humility/webinars</a></p> <p><b>Indigenous Cultural Safety Collaborative Learning Series:</b>  <a href="http://www.icscollaborative.com/webinars">http://www.icscollaborative.com/webinars</a></p> <p><b>Mr. Joe Gallagher, CEO First Nations Health Authority of BC: <i>Bringing Cultural Safety and Humility to Medical Practice</i></b>, Plenary session for the College of Physicians and Surgeons of BC Education Day, 14 September 2018.  <a href="https://www.youtube.com/watch?v=a1B2rPYbNKg">https://www.youtube.com/watch?v=a1B2rPYbNKg</a></p>	<p>Webinars to encourage learning about Indigenous cultural safety, self-reflection and positive change amongst healthcare professionals.</p>	
<p><b>Research Ethics and Guidelines</b></p>	<p><b>Description</b></p>	<p><b>Required?</b></p>
<p><b>TCPS2 Policy Statement:</b></p> <p>Canadian Institutes of Health Research, Natural Sciences and Engineering Research Council of Canada, and Social Sciences and Humanities Research Council of Canada,  <b>Tri-Council Policy Statement: Ethical Conduct for Research Involving Humans</b>, December 2018.  <a href="http://pre.ethics.gc.ca/eng/documents/tcps2-2018-en-interactive-final.pdf">http://pre.ethics.gc.ca/eng/documents/tcps2-2018-en-interactive-final.pdf</a></p> <p><u>Online TCPS2 tutorial:</u>  <a href="https://tcps2core.ca/welcome">https://tcps2core.ca/welcome</a></p>	<p>Core ethical policies for carrying out research involving humans. <b>*Chapter 9 is focused on research involving Indigenous Peoples*</b></p> <p>Reading the <b>policy document</b> is <u>required</u>.</p> <p>In addition, your REB may require research team members to successfully complete the online TCPS2 tutorial as part of the REB approval process.</p>	<p>✓</p>

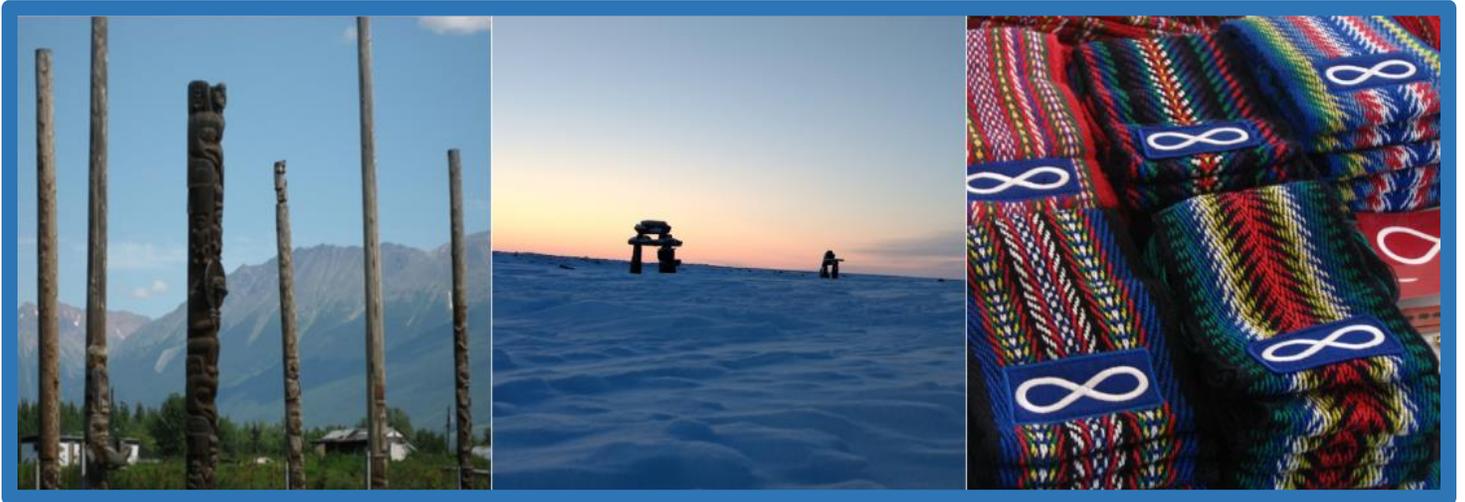
<p><b>First Nations OCAP Principles:</b></p> <p>The First Nations Information Governance Centre. <b>Ownership, Control, Access and Possession (OCAP™): The Path to First Nations Information Governance.</b> May 2014.  <a href="https://fnigc.ca/sites/default/files/docs/ocap_path_to_fn_information_governance_en_final.pdf">https://fnigc.ca/sites/default/files/docs/ocap_path_to_fn_information_governance_en_final.pdf</a></p> <p><b>OCAP website:</b> <a href="https://fnigc.ca/ocapr.html">https://fnigc.ca/ocapr.html</a>  <b>Video:</b> <a href="https://www.youtube.com/watch?v=SNpZw3DI4S4">https://www.youtube.com/watch?v=SNpZw3DI4S4</a></p>	<p>Set of standards of how First Nations data should be collected, protected, used and shared.</p> <p>Reading <b>pages 4-12</b> of the document is <u>required</u>.  Additional website and video options are suggested.</p>	
<p><b>DNA on Loan:</b></p> <p>Arbour L and Cook D. <b>DNA on Loan: Issues to Consider When Carrying out Genetic Research with Aboriginal Families and Communities.</b> <i>Community Genetics</i>. 2006, 9:153–160.  <a href="https://www.uvic.ca/medsci/assets/docs/other/DNA_on_loan.pdf">https://www.uvic.ca/medsci/assets/docs/other/DNA_on_loan.pdf</a></p>	<p>Guides how DNA from Indigenous research participants should be collected, stored, and used. DNA is ‘on loan’ to researchers rather than owned by researchers.</p>	
<p><b>International Guidelines:</b></p> <p>Taniguchi, NK, Taulii, M, and Maddock, J. <b>A comparative analysis of Indigenous research guidelines to inform genomic research in Indigenous communities.</b> <i>The International Indigenous Policy Journal</i>. 2012, 3(1).  <a href="https://ir.lib.uwo.ca/cgi/viewcontent.cgi?article=1069&amp;context=iipj">https://ir.lib.uwo.ca/cgi/viewcontent.cgi?article=1069&amp;context=iipj</a></p>	<p>A comparison of International guidelines for carrying out research in Indigenous communities.</p>	
<p><b>History of Harmful Research in Indigenous Communities:</b></p>	<p><b>Description</b></p>	<p><b>Required?</b></p>
<p><b>Indian (Havasupai) Tribe Wins Fight to Limit Research of Its DNA</b> - THE NEW YORK TIMES, 21 April 2010.  <a href="https://archive.nytimes.com/www.nytimes.com/2010/04/22/us/22dna.html">https://archive.nytimes.com/www.nytimes.com/2010/04/22/us/22dna.html</a></p>	<p>Media report of the misuse of Havasupai DNA for unapproved genetic studies, and the resulting lawsuit.</p>	

<p><b>Genetic Researcher Uses Nuu-Chah-nulth Blood for Unapproved Studies in Genetic Anthropology</b>- HA-SHILTH-SAH NEWSPAPER, 21 Sept 2000.  <a href="https://hashilthsa.com/archive/news/2013-07-22/genetic-researcher-uses-nuu-chah-nulth-blood-unapproved-studies-genetic-anth">https://hashilthsa.com/archive/news/2013-07-22/genetic-researcher-uses-nuu-chah-nulth-blood-unapproved-studies-genetic-anth</a></p> <p><b>Nuu-chah-nulth Blood Returns to West Coast</b> – HA-SHILTH-SAH NEWSPAPER, 16 Dec 2004.  <a href="https://issuu.com/hashilthsa/docs/25_december_16_2004">https://issuu.com/hashilthsa/docs/25_december_16_2004</a></p>	<p>Media reports of the misuse of Nuu-Chah-nulth DNA for unapproved genetic studies, and the return of blood samples to the communities. This case led to development of ‘DNA on Loan’ principles.</p>	
<p>MacDonald NG, Stanwick R and Lynk, A. <b>Canada’s shameful history of nutrition research on residential school children: the need for strong medical ethics in Aboriginal health research</b> (editorial). <i>Paediatric Child Health</i>. 2014, 19(2):64.  <a href="https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3941673/pdf/pch19064.pdf">https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3941673/pdf/pch19064.pdf</a></p> <p>Owens, B. <b>Canada used hungry Indigenous children to study malnutrition</b>. <i>Nature News</i>, 23 July 2013.  <a href="https://www.nature.com/news/canada-used-hungry-indigenous-children-to-study-malnutrition-1.13425">https://www.nature.com/news/canada-used-hungry-indigenous-children-to-study-malnutrition-1.13425</a></p> <p><b>Sick Kids Website Must Reflect Immoral Research on Indigenous Children</b>, THE STAR 25 July 2018.  <a href="https://www.thestar.com/opinion/contributors/2018/07/22/sick-kids-website-must-reflect-immortal-research-on-indigenous-children.html">https://www.thestar.com/opinion/contributors/2018/07/22/sick-kids-website-must-reflect-immortal-research-on-indigenous-children.html</a></p>	<p>Articles and media reports on the immoral nutritional experiments Canadian government scientists carried out on Indigenous children in the 1940s-1950s.</p>	
<p><b>Books</b></p>	<p><b>Description</b></p>	<p><b>Required?</b></p>
<p>CBC Radio: <b>Indigenous authors recommend books all Canadians should read (booklist on website)</b>:  <a href="https://www.cbc.ca/radio/unreserved/how-indigenous-authors-are-claiming-space-in-the-canlit-scene-1.4573996/indigenous-authors-recommend-books-all-canadians-should-read-1.4575751">https://www.cbc.ca/radio/unreserved/how-indigenous-authors-are-claiming-space-in-the-canlit-scene-1.4573996/indigenous-authors-recommend-books-all-canadians-should-read-1.4575751</a></p> <p>Gray Smith, M. <b>Tilly: A story of hope and resilience</b>. SONO NIS PRESS. February 1 2014.</p>	<p>A variety of fictional and non-fictional books written by Indigenous authors, and/or about the history of colonialism and Indigenous Peoples in Canada.</p>	

<p>Joseph, B. <b>21 Things you may not know about the Indian Act: Helping Canadians make reconciliation with Indigenous peoples a reality.</b> Page Two Books, Inc. April 2018.</p> <p>King, T. <b>The inconvenient truth: A curious account of Native people in North America.</b> Doubleday Canada, 2012.</p> <p>Lux, M. <b>Separate beds: A history of Indian hospitals in Canada, 1920s-1980s.</b> University of Toronto Press, Scholarly Publishing Division; 1 edition. March 14 2016.</p> <p>Sellars, B. <b>Price paid: The fight for First Nations survival.</b> Talonbooks. September 5, 2016.</p> <p>Sellars, B. <b>They called me number one: secrets and survival at an Indian residential school.</b> Talonbooks. April 15, 2012.</p> <p>Sterling, S. <b>My name is Seepetza.</b> Groundwood Books. November 1, 1992.</p> <p>Talaga, T. <b>Seven fallen feathers: Racism, death and hard truths in a northern city.</b> House of Anansi Press. September 30, 2017.</p> <p>Wagamese, R. <b>Indian horse.</b> Douglas &amp; McIntyre. January 27, 2012.  Film adaptation, premiered in 2017: <a href="http://www.indianhorse.ca/">http://www.indianhorse.ca/</a></p> <p>Weatherford, J. <b>Indian Givers: How the Indians of the Americas transformed the world.</b> Ballantine Books. November 29, 1989.</p>		
<b>Other</b>	<b>Description</b>	<b>Required?</b>
<p><b>Government of Canada website – Indigenous Peoples and Communities</b>  <a href="https://www.rcaanc-cirnac.gc.ca/eng/1100100013785/1529102490303">https://www.rcaanc-cirnac.gc.ca/eng/1100100013785/1529102490303</a></p>	<p>Information, maps, and statistics about Indigenous Peoples and Indigenous initiatives in Canada.</p>	

<p><b>United Nations Declaration of the Rights of Indigenous Peoples (2007)</b>  <a href="https://www.un.org/esa/socdev/unpfii/documents/DRIPS_en.pdf">https://www.un.org/esa/socdev/unpfii/documents/DRIPS_en.pdf</a></p>	<p>Describes individual and collective rights of Indigenous peoples around the world. It offers guidance on cooperative relationships with Indigenous peoples to states, the United Nations, and other international organizations based on the principles of equality, partnership, good faith and mutual respect.</p>	
<p><b>American Indian and Alaska Natives Genetics Resource Center:</b>  <a href="http://genetics.ncai.org">http://genetics.ncai.org</a>          Accessed 11 October 2018.</p>	<p>Resource guide to provide tribes and Indigenous peoples with the tools to make informed decisions about genetic research.</p>	

## PART 2: GENETIC COUNSELLING FOR *PRECISION DIAGNOSIS* STUDY



## PART 2: GENETIC COUNSELLING FOR *PRECISION DIAGNOSIS* STUDY

### I. GUIDING PRINCIPLES FOR GENETIC COUNSELLING

Given the long history of systemic racism and oppression, damaging government policies and institutions, and harmful relationships with unethical researchers and academics, it is imperative for Genetic Counsellors working with Indigenous families to have a deep understanding of the larger social and historical context surrounding their counselling interactions and how this may affect the patient-practitioner relationship and patient care. Genetic Counsellors must be mindful of the trepidation some patients may feel when entering a health facility or research study, demonstrate empathy and understanding, and be guided by the following principles that will help establish trust and build mutually respectful partnerships.

**Table 3. Guiding Principles for Genetic Counsellors to establish trust and build genuine, respectful partnerships with Indigenous clients.**

Guiding Principles	What they look like in practice:
<b>Transparency</b>	Open and honest engagement at all times; presenting all information needed for decision-making; not hiding any policies, protocols or information that patients would want to know; addressing all questions; admitting your limitations and being honest if you do not know something.
<b>Humility</b>	Being humble and eager to listen and learn from others; seeing yourself as a life-long learner; acknowledging that you are not always right; knowing that you are no more important than anyone else; integrating self-reflection into your practice.
<b>Partnership</b>	Minimizing the power differentials inherent in healthcare delivery; building respectful patient-practitioner relationships in which power and learning is shared. Patients are not passive receivers but rather powerful players in the relationship.
<b>Flexibility</b>	Having an open mind and the ability to build partnerships with those with differing world views, beliefs and values; ability to think outside of the box and problem-solve effectively when encountering new situations or hurdles.

<b>Self-determination</b>	Facilitating patient-centred care; ensuring that patients can make free and informed choices; valuing Indigenous Peoples' self-determination, as described by the <i>Ownership Control Access and Possession (OCAP)</i> Principles; allowing as much time as needed for decision-making (no pressure)
<b>Accountability</b>	Taking responsibility for your actions; being answerable to your patients, partners and stakeholders; doing what you say you will do; owning and communicating any mistakes you may make.

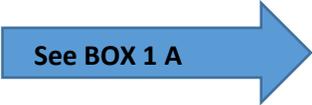
## II. POTENTIAL GENETIC COUNSELLING CHALLENGES

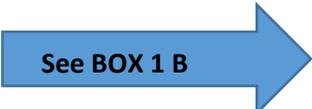
This section looks at specific areas of the Genetic Counselling interaction that may prove challenging, and provides ideas for building better partnerships with Indigenous clients.

The term 'challenges' is used in the context of the level of cultural safety experience and education a Genetic Counsellor has working with Indigenous Peoples and communities. Not all Genetic Counsellors will encounter the challenges below, but it helps to be prepared.

**Table 4. Common challenges in Genetic Counselling and tips for working through them.**

Challenges:	What Helps?
<p><b>Building Trust and Rapport</b></p> <p>Sharing of information and building relationships is tied to TRUST. Due to the history of broken promises and harmful experiences with healthcare providers and researchers, it may be more difficult to establish trust with Indigenous clients. It will likely take LONGER to build rapport.</p>	<ul style="list-style-type: none"> <li>➤ Have knowledge of our colonial history and resulting barriers...Be aware of experiences your client may be walking in with.</li> <li>➤ Take time! Accept a slower pace, do not pressure. Allow a session to go longer if needed.</li> <li>➤ Contract longer at the beginning of session.</li> <li>➤ Become comfortable with silence. Do not interpret a slower pace or silence as ineffectiveness.</li> <li>➤ Listen more and talk less. Try to ensure the client has finished speaking before you contribute to the conversation.</li> </ul>

<p><b>Assessing Pedigrees</b></p> <p>Clarifying biological vs non-biological relationships can be challenging due to wide definition of ‘family’, fluid guardianship, the disproportionate number of Indigenous children in foster care, and the fracturing of families due to the impacts of colonization.</p> <p>Asking family history information often inadvertently bring the history of family deaths/suicides, residential schools, 60’s scoop, Indian hospitals, family separations, and other traumatic events to the surface.</p>	<ul style="list-style-type: none"> <li>➤ At the start, spend more time explaining the background reason for pedigree-taking and why we emphasize biological relationships</li> <li>➤ Plan more time to collect pedigree information</li> <li>➤ Be prepared to clarify biological vs non-biological relationships –e.g. adoptions within and between families may be common, terms like ‘auntie’, ‘uncle’ and ‘cousin’ are often used to describe various relationships (including non-biological), families may not consider it important to distinguish full-sibs from half-sibs</li> <li>➤ Be sensitive and tread lightly. Emphasize at the beginning that client can ask you to stop at any time if they feel uncomfortable.</li> </ul>
<p><b>Logistical Barriers</b></p> <p>Many logistical barriers to following the study protocol or providing care may be encountered, including: long distance travel for rural clients to access services; lack of transportation; lack of health/counselling services in local community; burden of personal out-of-pocket expenses for healthcare or travel; difficulty contacting clients.</p>	<ul style="list-style-type: none"> <li>➤ Be flexible and think outside of the box – is there another way something can be accomplished?</li> <li>➤ Be an advocate – help your client problem-solve so they can access health services.</li> <li>➤ Seek help – build connections with agencies, community support workers, local health centres and Indigenous patient liaisons who can advocate on behalf of your client.</li> </ul>
<p><b>Collective Decision-Making</b></p> <p>Drawing strength from family and community. Indigenous clients may be more likely to involve family members or other community members in decision-making.</p>	<ul style="list-style-type: none"> <li>➤ Support the inclusion of individuals your client chooses to involve in decision-making, but also check-in with your client individually.</li> <li>➤ Be open to liaising with other family members, if requested by your client and if they give consent to share information.</li> </ul> <div style="text-align: right; margin-top: 10px;">  </div>

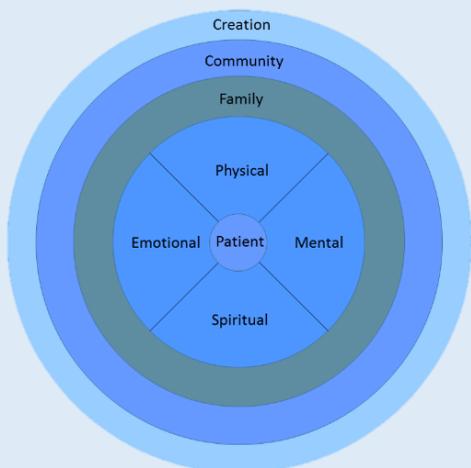
<p><b>Differing Views on Health and Wellness</b></p> <p>Clients may have different world views and beliefs about spirituality, disease etiology, use of traditional medicine/healing practices</p>	<ul style="list-style-type: none"> <li>➤ Be open to different ideas of ‘wellness’.</li> <li>➤ Recognize the Western medicine bias of our profession.</li> <li>➤ Ask about clients’ own views of wellness and coping strategies. <u>Do not assume</u> everybody uses traditional healing practices.</li> <li>➤ Consider the concept of ‘<b>two eyed seeing</b>’<sup>1</sup>, which may apply in some situations. Recognize that Western medicine and Indigenous ways of knowing, including traditional healing practices, can usually work together and are not mutually exclusive (see Box 1,C).</li> </ul> <p style="text-align: right;"><b>See BOX 1 B-C</b> </p>
<p><b>Recognizing Importance of Spirituality in Health</b></p> <p>Many, but not all, Indigenous families have spiritual beliefs and practices that are integrated into all aspects of life, and are important for health, coping, and well-being.</p>	<ul style="list-style-type: none"> <li>➤ Studies indicate that Genetic Counsellors may be <u>less</u> religious/spiritual than the general population, and often do not incorporate spiritual assessment into their counselling sessions.<sup>2,3</sup></li> <li>➤ In open-ended interviews, 8 out of 10 Gitxsan women identified spirituality as a major factor facilitating their coping with a diagnosis of Long QT syndrome in their family.<sup>4</sup></li> <li>➤ Do not assume all Indigenous clients are ‘spiritual’, but work at getting more comfortable asking if and how clients’ spiritual views may relate to health and genetics.</li> </ul> <p style="text-align: right;"><b>See BOX 1 B</b> </p>

**Box 1. Examples of Indigenous Ways of Knowing That May Contribute to Client’s Views on Health and Well-Being and Influence Healthcare Approaches.**

**A) Common differences between Western and Indigenous values and beliefs.<sup>4</sup>**

Western Cultural Values & Beliefs	Indigenous Cultural Values & Beliefs
Individual autonomy	Sociocentric (connection to family, community, natural & spirit world)
Individualism	Collectivism
Linear	Circular

**B) A holistic view of health is common for Indigenous Peoples. Some First Nations use the concept of the *Medicine Wheel* to visually depict the layers and connections that contribute to the health of individuals and communities.** Medicine Wheel Image from: Huisman. L (2010).<sup>4</sup>



*“The Medicine Wheel concept from Native American culture provides a model for who we are as individuals. We have an intellectual self, a spiritual self, an emotional self, and a physical self. Strength and balance in all quadrants of the Medicine Wheel can produce a strong, positive sense of wellbeing, whereas imbalance in one or more quadrants can cause symptoms of illness. Addressing issues of imbalance can potentially diminish your patient’s symptoms and enrich their quality of life.”*  
 (Montour, 2000)<sup>5</sup>

**C) The concept of ‘Two Eyed Seeing’,<sup>1</sup> conceived by Mi’kmaw elder Albert Marshall, depicts the strong partnership, humility, and respect for differing views that is necessary for the provision of culturally safe healthcare.**

**‘Two Eyed Seeing’**  
 Mi’kmaw elder Albert Marshall

To see from one eye with the strengths of **Indigenous** ways of knowing

and to see from the other eye with the strengths of **Western** ways of knowing

And to use **both** of these eyes together

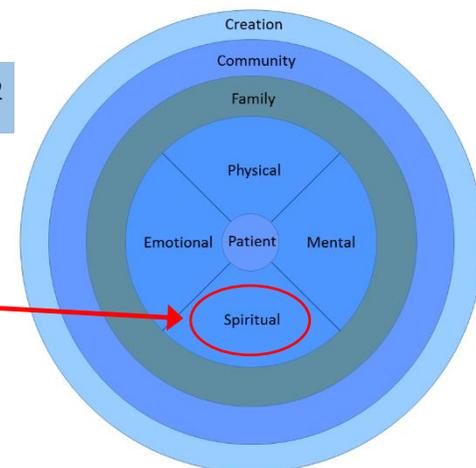
### III. DETAILED GENETIC COUNSELLING PLAN FOR *PRECISION DIAGNOSIS* ENROLMENT SESSION (v.2, 21 August 2019)

#### Guiding Principles:

- Remember the principles of **Transparency, Humility, Partnership, Flexibility, Self-Determination, and Accountability** that should guide each session. Refer to Table 3, p.18-19 of Genetic Counselling Guide
- Always be respectful and act genuinely in your interactions and relationships.
- Be aware that healthcare interactions can trigger strong emotions associated with the negative history of colonization, residential schools, 'Indian hospitals', and child welfare system. Acknowledge that racism and discrimination continue to impact Indigenous individuals and communities today.
- Take your time! Do not pressure participants to make decisions quickly. Be sure to book enough time for the session so it is not rushed.

#### Contracting:

- More time may be needed for contracting at the beginning of the session to lay the groundwork for building trust/rapport, create an open and safe space, assess views of health and wellness, and develop a plan for the session that meets both genetic counsellor's and client's goals. Refer to Table 4, p.19-21 and Box 1, p.22
- Indigenous clients often have a holistic view of health and well-being. Remember to probe for if the participant has any spiritual beliefs related to health/genetics.



#### Taking Pedigrees:

- At the start, spend more time explaining the background reason for pedigree-taking and why it is important to clarify biological vs non-biological relationships for genetic testing purposes. Refer to Table 4, p.20
- Be sensitive and tread lightly. Family history questions may bring up past traumas. Explain at the beginning that clients can ask you to stop at any time if they feel uncomfortable.
- Remember to ask targeted family history screening questions r.e. the particular disorder in the family, as well as cardiac disorders and cancer (conditions where Incidental Findings commonly arise). Please refer to the *Genetic Counselling Appointment Checklist* included in the appendix for reminders of family history screening questions.

## Study Information to cover during enrolment session:

### Basic Genetics

See NSGC **Genomic Counselling Guide**<sup>6</sup> for helpful visual aids

- Cells, chromosomes, genes, variants
  - May be useful to use analogies such as: genome = book, chromosome = chapter, gene = sentence, base pair = letter, variant = different spelling
- What are variants?
  - Everyone has variation in their genome. Some variants have no effect on gene function and health, while some disrupt gene function and cause health conditions. Sometimes we do not have enough information to tell us if a certain variant contributes to a health condition or not (i.e. Variant of Uncertain Significance – VUS)
  - Review that variants can have variable expressivity and penetrance.

### Whole Genome Sequencing (WGS) Technology

- The participant may have already had some genetic testing (e.g. a few genes have been sequenced or a test for CNVs has been done)
  - With WGS we are now taking a closer and un-biased look at the *entire genome all at once* to see if we can find any variants in the genome which are possibly related to the primary diagnosis
- Participants' **DNA is considered "on loan"** for the purposes of this research. We consider research participants to be the owners of their DNA and other biological samples. Participants can request that samples be destroyed or sent back to them at any time, with written confirmation that this has been done.<sup>7</sup>



### The Process

- DNA Sample collection
  - Blood is the preferred way to obtain DNA for WGS. Inform participants of the most convenient locations to get blood drawn.
  - In cases where there are significant barriers to blood draw (e.g. access or needle anxiety), LifeLabs can arrange to send a buccal or saliva kit to participants for DNA collection. Buccal collection is preferred over saliva.
- DNA extraction
  - Once the participant's sample is collected, it is sent to LifeLabs in Toronto for DNA extraction.
  - LifeLabs will not keep any DNA, but will send it to the Genome Sciences Centre (GSC) in Vancouver for WGS.
- Whole genome sequencing
  - Participant's DNA will be sent to the GSC in Vancouver where WGS will be carried out.

- Bioinformatics
  - Our research team will analyze the data generated from WGS.
- Preliminary Results
  - Our research team will discuss initial findings and identify any parts of the participant’s DNA that may be of interest. As noted below, in cases where a possible disease-causing variant is identified, DNA samples will be sent back to LifeLabs in Toronto for clinical confirmation of the variant.

### Possible Results and Their Implications

- Possible results fall under three broad categories:
  - 1) **Possible disease-causing variant**
  - 2) **No reportable variants (i.e. no ‘possible disease-causing’ variants found)**
  - 3) **Incidental Findings**

- If a **possible disease-causing variant** is identified:
  - Such results must be technically validated by a clinical-grade test (e.g. Sanger sequencing, MLPA) at LifeLabs to ensure they are true findings, before being reported back to referring doctor. At this point LifeLabs will issue a clinical report, which becomes part of the participant’s medical record.
  - In the process of clinical reporting, LifeLabs will classify variants according to ACMG guidelines.<sup>8</sup> We expect most results will be classified as either ‘likely pathogenic’ or ‘pathogenic’, but it is possible that some variants may be classified as VUS, even if our team believes there is compelling evidence for disease causation.
  - Such results may provide important health information for proband, siblings, extended family, and may provide reproductive information.
  - Such results may provide treatment and/or surveillance options.
  - We estimate an approximate 30% diagnostic rate overall, i.e. 30% chance of finding an answer.
- If **no reportable variants are** identified: this “negative” result may indicate several possibilities:
  - 1) The cause is not monogenic.
  - 2) The cause is monogenic, but we failed to identify it for technical reasons.
  - 3) The cause is monogenic, but in a gene not yet recognized as causing disease.

*If no reportable variants are identified, there is still a chance that re-analysis throughout the study duration may yield a diagnostic finding.*

#### How will we handle Variants of Uncertain Significance (VUS)?

**VUS** = a gene variant with uncertain clinical significance. There is not enough information to determine whether it is related to disease or not.

- In most cases where our study team identifies a variant believed to be a VUS, this variant will not be forwarded for clinical confirmation but will fall under the category of “**No reportable variants**”.
- In certain cases where our study team believes a potential VUS has compelling pathogenic-like features and is in a gene with a strong match to the proband’s phenotype, this variant may be forwarded for clinical confirmation. Even if this finding is classified by LifeLabs as a VUS, it will be considered a **compelling VUS** and will still be returned to the family.
  - There may be the option of further testing to help interpret a VUS result (e.g. functional studies or segregation studies)

- **Incidental Findings (IF):** variants known to cause/increase risk for a disorder which is unrelated to the condition the patient/family was being tested for.
  - We are not specifically screening for these types of results, but we anticipate an IF will be found in ~ 3% of study participants.
  - IFs that **will** be reported (consistent with current CCMG guidelines)<sup>9</sup>:
    - In a child participant, we will report any childhood onset, medically actionable conditions (e.g. childhood onset hereditary cancer, Long QT, neurofibromatosis). Although we will not usually report back any IFs causing adult-onset conditions in children, we will report *some* adult-onset risk alleles if we strongly believe it is in the child's or family's best interest (e.g. highly penetrant hereditary adult-onset cancers where a possibly affected parent was not enrolled).
    - In incompetent adults, we will report back any medically actionable IFs.
    - Competent adult participants have the choice of whether or not they wish to be informed of any medically actionable IFs discovered in them.
  - IFs that **will not** be reported:
    - Non-actionable findings (e.g. variants causing Alzheimer disease, finding that one parent is a carrier of an autosomal recessive condition), non-paternity, incidental variants of low penetrance (e.g APOE4, pharmacogenomic variants [exceptions for pharmacogenomic variants could be made when circumstances of applicability for a particular participant are already known to the research team]).

### Limitations of Study:

- It is important to understand the limitations of the WGS performed for this study. This study focuses on finding disease-causing gene variants *related to the specific condition* in the affected person in your family, and not on finding gene variants that may cause other *unrelated* diseases. In other words, the analysis of each genome is guided by the health problems in the family that we are aware of and are actively investigating. Therefore, **the WGS performed for this study is not a comprehensive analysis of all genes, and a 'negative' result does not mean the absence all of genetic risk factors.**
- Knowledge about genes is rapidly changing, such that what we think we know now about a participant's genes may be shown to be incomplete, or even inaccurate, in future years. Hence, it is important to consider re-connecting with a geneticist or genetic counsellor when making medical decisions based on a genetic diagnosis. In addition, whether the participant receives positive or negative results from this study, we may get more information about their result over the course of the study.

### Possible Harms of Undergoing WGS

- Discomfort of blood draw
  - *Note that while buccal or saliva samples are an option, whole genome libraries from these samples are generally of inferior quality compared to those from blood. This is largely due to presence of contaminating microorganisms in the oral cavity as these are also sequenced. Consequently, fewer reads map back to the human genome. Reduced DNA yield is also expected from buccal swabs and saliva compared to blood as the DNA is likely to be degraded by the*

*enzymes in the mouth. You can explain that this is why blood is preferred if your participants are trying to decide between blood and buccal/saliva.*

- Potential psychological impact
  - Discovering a new condition in the family may change life planning and decisions – some people just prefer *not to know*
  - Some genetic conditions discovered through WGS are inherited, and are present in other blood relatives as well. Therefore, genetic information can potentially impact the wider family, not just the individuals participating in the study.
- Potential risk of disclosure of your genetic information. Despite our best efforts to protect your privacy, complete anonymity cannot be guaranteed.
  - If a pathogenic or possibly pathogenic variant (including IF) is identified, it will be clinically confirmed by LifeLabs before the result is reported back to referring doctor. LifeLabs will issue a clinical report and, at that point, **the results will become part of the participant's medical record**, accessible to other healthcare providers involved in the participant's care. This removes the option of participants keeping their result completely private and out of their medical record.
- Potential risk of genetic discrimination, despite the *Genetic Non-Discrimination Act (GNA)* currently in place.
  - Offer to provide participants with the CAGC Fact sheet on the GNA.<sup>10</sup>
  - Participants have no obligation to share their genetic test result with insurers or employers, despite some insurance organizations asking people to voluntarily disclose this information.

#### **Possible Benefits of Undergoing WGS**

- Participants may receive an explanation for the condition in their family.
- The genetic finding may lead to a diagnosis, which may lead to more information on anticipated outcomes, better informed care, access to resources, empowerment, grieving/acceptance, information about recurrence risk, and a way to explain the condition to family and friends.
- Information discovered through this study may benefit others with suspected genetic conditions.

#### **Turnaround Time**

- It is difficult to predict how long results will take to come back but we estimate approximately 6 months.

#### **Reporting of Results**

- **Possible disease-causing variants** (including IFs, where applicable) will be returned to the referring physician who will contact the family for an appointment. The genetic counsellor will join this appointment, or if not possible, follow up with a phone call.
- If **no reportable variants** are found, the family may either receive this information from their referring physician or may receive a phone call from the study genetic counsellor to discuss this. The genetic counsellor will also write a study results letter back to the referring physician.

#### **Follow-up**

- Throughout the length of the study (~4 years) we will likely re-test participants' DNA samples and/or re-analyze their data as our knowledge and WGS capabilities grow. Therefore, it is possible that participants may receive new gene variant results at a later date, or the interpretation of their results

may change over the course of the study (i.e. variant classification may be 'upgraded' or 'downgraded' over time due to improved technology and knowledge).

- With some results, there may be the possibility of doing further studies to understand the full nature of the condition in the family (e.g. functional studies, SOLVE-RD) – if this is the case, and study team thinks additional studies could be helpful, we will approach the family for separate consent.
- There is also an optional consent tick-box included on the main study consent form to be re-contacted regarding future research studies (separate from this study).
- Please call or e-mail us at any time if you have any questions about the study.

## Informed Consent

### Written Consent

- Consent/assent forms are to be reviewed with study participants and the encounters clearly documented in the research chart. It is important to document if participants are capable of understanding the consent/assent and if they show agreement. If it is determined that it is inappropriate to obtain assent from a participant (like a child or incompetent adult), it is important to document this and the reasons why.

The applicable consent/assent forms are to be signed by families who agree to join study:

- Authorization to Release Healthcare Information (ROI)
- Main study consent form - for adult participants and parent/guardians of child participants
  - Including optional consent tick-boxes for photos, IFs, and future re-contact
- Adolescent assent form - generally for participants aged 14-18 years
  - Including optional consent tick-boxes for photos and future re-contact
- Child assent form - generally for participants aged 7-13 years
- LifeLabs requisition

### Notes on parental/guardian consent for children or incompetent adult participants

- When parents are the legal guardians of a child participant:** Although the consent form only requires the signature of one parent, it is recommended that both parents agree to their child's study participation. In cases where only one parent is present during the enrollment session and/or only one parent appears to be involved in the child's care, we suggest that the genetic counsellor discuss this recommendation with the study participants and document this discussion.
- When parents are not the legal guardians of a child participant:** For children who are not in their parents' care (for example, children in foster care, children with other legal guardians), it is important to confirm who the legal guardian is. Only the legal guardian can provide consent for study participation. In the case of foster care, the social worker is usually the person who can provide consent. Ideally, the child's biological parent(s) would also be aware of, and in agreement with study enrolment, since their participation in the study would be helpful and WGS results could have implications for their family.
- For incompetent adult participants:** It is important to confirm who their legal substitute decision-maker is. Only the legally authorized substitute decision-maker can provide consent for study participation.

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