# Genome-Wide Sequencing Counselling Guide

## Sequencing facilitated by TIGeR for Alberta Precision Laboratories

Your physician has recommended a genetic test known as **Genome-Wide Sequencing (GWS)** to identify the genetic cause for your or your child's condition. Genome-wide sequencing involves looking at genetic information in thousands of genes, searching for a genetic difference (variant) that could be the cause of the condition. The laboratory and the health professionals involved in your care will use information from your medical records, including your family history, to help interpret the results of this test.

**Potential benefits of the test:**

- To help explain your or your child's condition.
- To give more information about the diagnosis and what it means for you or your child.
- To give more information about what the condition means for other family members and any future pregnancy.

**Limitations of the test:**

- Although the purpose of the test is to find a genetic variant to explain your or your child's condition, sometimes results may be unclear. For example, genetic variants may be reported that are not yet fully understood (variants of uncertain significance) or found in genes that are not yet fully understood (genes of uncertain significance).
- This test will not identify all genetic variants. Some will be missed.
- A negative test does not exclude the possibility that there is an underlying genetic cause for your or your child's condition. Reanalysis may be an option in the future.
- Due to the growing knowledge in genetics, it is possible that the understanding and interpretation of your results may change over time. Therefore, please keep in touch with your physician.

**How the test is done:**

- This test will be performed on a biological sample (e.g., blood).
- Biological samples or clinical information from your biological parents and/or biological family members may be requested to help interpret the results.
- Results will be discussed with you and put in your medical record.

**Possible risks of the test:**

- Results may have implications for your future health and possibly for other family members.
- This test could also detect non-biological relationships, such as adoption or non-paternity.
- It is possible that results not related to your or your child's condition may also be found. Though we do not actively look for these results, we will share such results with you. This type of result will be considered an "incidental finding".
- Currently, in Canada, these results are unlikely to have an impact on your insurability or employment.

**Secondary findings:**

- Each person providing a sample for genome-wide sequencing (GWS) has the option of learning about "secondary findings". These genes may not be directly related to the reason for ordering the test, but they are known to be associated with a higher chance of certain serious health conditions. If you choose to have these genes analyzed, our laboratory will complete a targeted analysis of a recommended set of genes.
- Our laboratory follows guidelines issued by a professional association, the American College of

Medical Genetics and Genomics.

**Consent to Contact**

- Consent for research contact - GWS is a test that was developed to try to diagnose patients with very rare genetic conditions. Enabling research helps improve our understanding and treatment of such rare conditions. You have the option of being contacted for future research opportunities which have been approved by Research Ethics Boards.
- Consent for clinical data sharing - It is critically important that laboratories share data to improve test performance and ensure that we are providing the best possible test for your family and for other patients. With your consent, we will share your/your child's GWS coded data (information that can identify you will be replaced by a code) and clinical features with an approved Clinical Knowledge database(s) in Canada.

**Confidentiality**

- Results of GWS will only be reported to the health care provider(s) who ordered the test. The laboratory will not give test results to other individuals without the patient's written permission, or unless required by law. The written report will become part of the patient's medical record.
- With **consent**, coded GWS data (i.e., information that can identify the patient will be replaced by a code) and clinical features will be shared through institutionally approved Clinical Knowledge Bases within Canada. Coded data shared in Clinical Knowledge Database(s) can include demographic information (sex, age, and ethnicity), details of the patient's clinical presentation, diagnoses, and genetic variants. This data will only be accessible to professionals working in diagnostic laboratories in Canada.
- Only the laboratory where the patient's test is performed will have access to an individual patient's full data set.
- Leftover de-identified samples may be used for clinical laboratory validation and quality assurance.

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# Genome-Wide Sequencing Acknowledgement and Consent

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By signing this form, you acknowledge that you wish to proceed with Genome-Wide Sequencing.

**Secondary Findings**

You have the option of learning about secondary findings. These genes may not be directly related to the reason for ordering the test, but they are known to be associated with a higher chance of certain serious health conditions. If you choose to have these genes analyzed, our laboratory will complete a targeted analysis of a set of genes, as recommended by the American College of Medical Genetics and Genomics.

Do you consent to the reporting of secondary findings?

YES ☐ NO ☐

**Consent for Clinical Data Sharing**

It is critically important that laboratories share data to improve test performance and ensure that we are providing the best possible test for your family and for other patients. With your consent, we will share you/your child's GWS coded data (information that can identify your will be replaced by a code) and clinical features, as described in the counselling guide. You may withdraw from such contact at any time.

I consent to share my coded data with Clinical Knowledge Networks approved in Canada

YES ☐ NO ☐

**Consent for Future Research Contact**

You can provide permission to be contacted regarding future research opportunities relevant to you in the future. You may withdraw from such contact at any time.

Would you like to be contacted about future research opportunities for which you may be eligible?

YES ☐ NO ☐

| **Patient Signature** | |
|---|---|
| Name: | Date of Birth: |
| Signature: | Date of Signature: |
| Name of Parent or Guardian (if applicable): | Relationship to Patient: |
| Phone number: | Email address: |