Exome Sequencing

Information for Patients

What is exome sequencing?

Exome sequencing is a test that can help diagnose genetic conditions.

- Genes are instructions for the cells of your body.
- Changes in genes can lead to a variety of symptoms or health conditions that may run in families.
- Exome sequencing looks at all your genes to search for changes that might explain why you have a symptom or condition.
- Exome sequencing is usually done on a small amount of blood.
- Samples from your blood relatives may also be requested (for example, a child and both of their biological parents) to help understand the results.

What are the steps?

- Talk to your clinician and decide together if exome sequencing is right for you
- Speak with your clinician about what samples are needed from your family.
- Results will take several weeks to months.
- Follow up with your clinician to discuss the results of your genetic test.
- You may be referred to other health care providers for follow up depending on results.
- In the future, your clinician may be able to re-examine your exome. Connect with your clinician to understand if or when this may be an option.

What are the possible results?

Your exome genetic test result may:

1. Show one or more gene changes that explain your health condition.
2. Show no reported gene change to explain your health condition.
3. Show a gene change that is not yet fully understood and may or may not explain your condition (variant of uncertain significance).
4. Show a gene change that does not explain your current condition but can impact your health (secondary finding). More tests or treatments may be suggested.
   - For example, an increased risk of a health condition like cancer or heart disease.
   - In some cases, you can choose whether you want to receive secondary findings. Talk to your clinician about your options.
5. Show an unexpected result not related to your health condition.
   - For example, a parent is not a biological parent, or parents are related to each other by blood.

For more information

Visit the Genome-wide Sequencing Ontario website for more information:
www.gsontario.ca/for-patients/

Need this information in an accessible format? 1-877-280-8538, TTY 1-800-855-0511, info@ontariohealth.ca.

Document disponible en français en contactant info@ontariohealth.ca.

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