Genomic Testing Results

A guide to understanding your genomic testing results and exploring your next steps

*Some parts of this booklet have been filled in by your healthcare provider. These personalized sections are marked with a star (★). You will see some medical terms. These words are in **bold**, and explained with colour-coded definitions.*

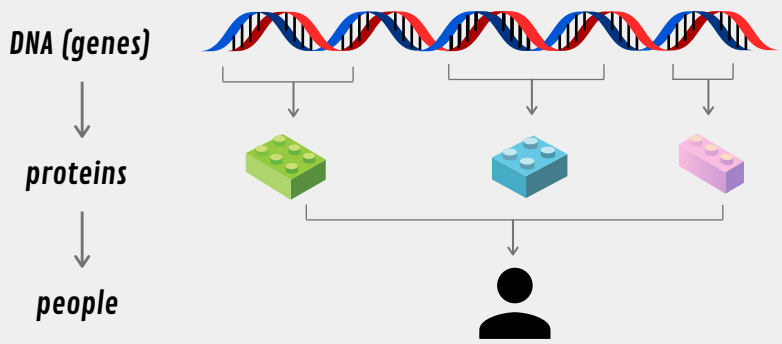
Background Genomic Information

Your **DNA** is the information inside your cells that is needed to build and maintain your body. Your unique set of DNA is called your **genome**. We still don't know the role of every part of the **genome**, but scientists are continuing to work to understand it.

Your **DNA** comes as a matched set: half of it comes from your mother and half from your father.

Some sections of your DNA make up **genes**, which are the instructions to build proteins.

Proteins are the building blocks of your body



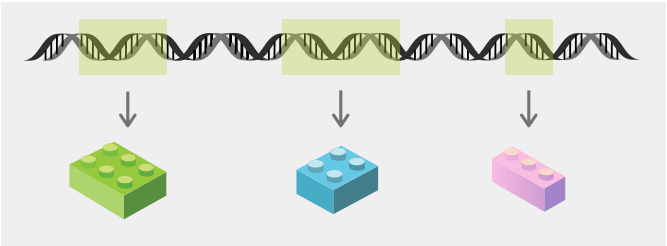
Differences in DNA are known as **variants** or mutations. They occur in everyone and most are perfectly normal. Sometimes a **variant** can change the instructions for making a **protein**, and this can cause a medical problem. This is what we were looking for in your genetic testing.

"Type of inheritance" describes the way **DNA** is passed from parent to child. Each **DNA variant** can be inherited from a mother or a father, or sometimes both. Other times, **variants** are not inherited from either parent; they may show up for the first time in the child. Some genetic conditions need two copies of a **variant**, one in each **gene** (like a double dose of a drug) to cause a genetic problem. Other genetic conditions need just one dose.

Remember, we have no control over our **DNA**, the **variants** we pass on, or the **variants** that show up in us for the first time. We cannot choose to pass down our **DNA** for things like eye or hair colour. Likewise, we cannot control whether we pass down potentially disease-causing **DNA**. The variation present in our **DNA** is what makes us unique, regardless of the outcomes of those **variants**.

*For more information on these topics and more, visit **MedinePlus – Help Me Understand Genetics***

Background Genomic Testing Information



In this image, the **DNA** highlighted in green is directly involved in making protein. This is called **protein-coding DNA**. There is still lots of DNA in-between. This is called **non-coding DNA**. **Non-coding DNA** can still impact how **proteins** are made. For example, it can work by speeding up or slowing down the production of **protein**.

Some **genomic** tests look at all of the **DNA,** while others look at just the **protein-coding DNA.**

Your Genomic Testing Results

Date:

Name:

★ ***The genomic testing you received is called:*** **Whole Exome Sequencing (WES)**

Whole Exome Sequencing (WES) only reads the part of your genome that codes for proteins (protein-coding DNA). Most of the variants we know about now are in this portion of the DNA, so this test will find the majority of disease-causing variants that are currently known.

★ ***Who was tested?*** **Trio**

The person with the medical condition and their biological parents were tested.

★ ***Your testing results:*** ***There has been a finding linked to your medical condition.***

Here are some more details about the **DNA variant(s)** that have been identified:

Affected gene name(s):

What does this gene do in our bodies?

Variant(s):

Are there known medical conditions that may be caused by changes in this gene?

Inheritance of your variant(s): **De novo (Dominant)**

This is a type that appears for the first time in a child.

Your Genomic Testing Results

Genomic testing results can be complex and hard to understand. You might receive both lab results and a doctor's interpretation, which might seem different. The lab looks for **variants** in **DNA**, and classifies them using computer tools and comparison's to other people's **DNA**. Your doctor then figures out if the **variant** could be causing the medical problem that you or a family member has. Sometimes, nobody can be certain what a particular **variant** in the **DNA** means.

★ ***The lab interpretation is that your variant is* a Variant of Uncertain Significance (VUS)**

The lab found a difference in the DNA, but not enough is known about the variant to say whether it might be causing the concerns.

Remember, the lab interpretation is not based on your doctor's interpretation of the results.

★ ***Based on your symptoms, your doctor thinks that it is* very unlikely *that this variant is causing your condition. Here are some more notes from your healthcare provider:***

During future consults, different doctors may interpret these results in a different way. Each specialist may have unique suggestions for your health management.

★ ***Here are some details about Incidental (Secondary) Findings:***

While searching your **genome**, the lab sometimes finds **variants** that are not causing the problem being tested for, but might cause other possible health issues. These are called *Incidental (Secondary) Findings*. Before testing, you chose whether or not you wanted us to look for them.

There was an incidental finding. Your doctor will give you more information.

Geneticists are still learning to understand all of the information in our **genome**. This is why genetic testing results can often seem "fuzzy" or uncertain. As genetic testing technology improves, understanding of your **variants** might change. If you get an uncertain result, you can check back with your doctors from time to time to see if new information is available.

There is a lot of information to process. Take your time. Your next steps may include connecting with certain resources, visiting your family doctor, or searching for a specialist. If you need support with this process, or still have questions about your results, please contact a genetic counsellor or doctor.

Resources and Support

Learning about genetic conditions can be complicated and may take time. Many people need extra help. We know you have been putting in hard work for a long time. You may already be well connected to helpful resources. Here are some other tips, websites, and tools that may be useful after receiving **genomic** testing results. **Remember, you are not alone.**

When searching for resources, keep in mind that...

- Medical literature can be very technical and hard to read, and it often shows the worst symptoms. Patient websites are often more friendly when looking for information.
- Each **DNA variant** of a **gene** can cause different symptoms, which is why your family's medical issues can be quite different from another person with a **variant** in the same **gene**. Although it sounds like the same problem, it might not be. Focus on the diagnosis your doctor shared.

FindSupport BC: Powered by the *Family Support Institute of BC*, this website is meant to help families of persons with disabilities navigate the resources that are available to them. Make sure to look for the **Disability Alliance BC**, **Planned Lifetime Advocacy Network**, and **Inclusion BC**.

Rare Disease Foundation: This foundation has a *Family Counselling Assistance Program* and a *Parent 2 Parent* Resource Network. They have a good booklet called **Living Without a Diagnosis**. There is also the **Rare Disease Information and Support Line** which you can call to receive help navigating a rare disorder.

HealthVault, **Medical Records**, **myPHR, My Medical Free:** These smartphone apps allow you to track your medical history, store results from your medical tests, and share information with family or doctors. Make sure to check the security and privacy settings.

MyBooklet BC can help you design a personalized medical information booklet which summarizes your medical history in a way that is easy to share with health care providers and support workers.

Scientific Information Sources – **Orphanet** has information for doctors and researchers, so the language can be complex. It shows previous research and medical cases for a specific gene or diagnosis. Remember that different variants in a gene can produce different health conditions, so make sure that the information matches your exact testing results.

Click on the links above, or type in the website name to your search engine.

Your Next Steps

Navigating your **genomic** results is not something you have to do alone. It is your doctors' and/or genetic counsellors' job to make sure you understand what the **genomic** results mean and support you through your next steps. You can help them support you by telling them what information is most important to you.

Here are some questions patients in your position often ask their doctor or genetic counsellor:

What are these **DNA variants**? Which **genes** are affected? How do these **variants** impact my health? Where did these changes in the **DNA** come from? Were they inherited? How can we change our health management? Are there other doctors we should see? Can this result change over time?

Sharing your results is entirely up to you...

If you do choose to share your results, these are some people you may share them with.

- **Family:** Genetic information may be important for the health or family planning of relatives. Family and friends may be better able to help and support you if they can inform themselves about the genetic diagnosis.
- **Your healthcare team:** A genetic diagnosis can change health management or help your healthcare team understand your medical condition. Only some of your doctors have access to your genetic results. Others, such as therapists or caregivers, likely do not. If you are unsure how to explain your results to your doctors, you can share this booklet with them.
- **Online platforms and genetic networks:** There are websites that connect families to those with similar diagnoses. Posting information about your genetic results on some scientific databases can help to deepen scientists' understanding, and advance research.

Support groups allow people who have similar experiences, diagnosis, or symptoms to connect...

Make sure to check the privacy and security of each platform before sharing your information.

- **Facebook** has many local and international support groups.
- **Global Genes – Allies in Rare Diseases:** This is a rare diseases support network to connect the individuals in the rare disease community, find resources (**RARE List**), and other families with shared experiences (**RARE Portal**, **RARE Daily**, **RARE Cast**).
- **MyGene2**: Create an account to share your variants with other families, researchers, and clinicians. You can also search for families with variants of the same gene and control how much you share.
- ★ Recommended support groups for your variant or condition:

This booklet is designed to be a supplement for genetic counselling, not a replacement for it. You can find a genetic counsellor in your area by using the CAGC Find a Clinic tool, or the NSGC Find a Genetic Counsellor tool.