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.*

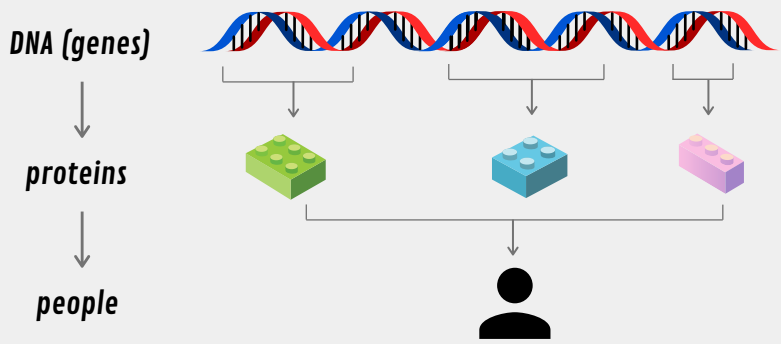
Background Genomic Information

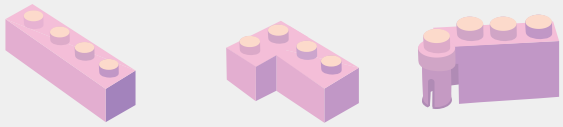
Your **DNA** is the information inside your cells that is needed to build and maintain your body. Your unique set of DNA is called your **genome**. We still don't know the role of every part of the **genome**, but scientists are continuing to work to understand it.

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





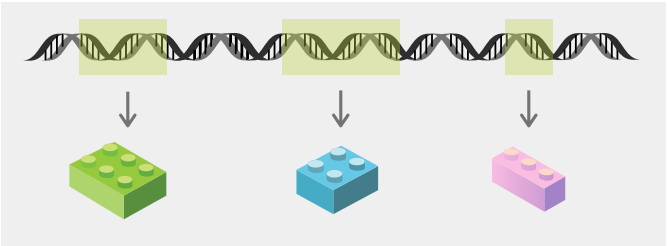
Differences in DNA are known as **variants** or mutations. They occur in everyone and most are perfectly normal. Sometimes a **variant** can change the instructions for making a **protein**, and this can cause a medical problem. This is what we were looking for in your genetic testing.

"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 Genome Sequencing (WGS)**

Whole Genome Sequencing (WGS) reads your entire genome. It can find variants in the DNA that makes up genes (protein-coding DNA) as well as variants in the DNA in-between (non-coding DNA). Currently, it is the most thorough genetic test available to look for variants that might cause a medical concern.

★ ***Who was tested?*** **Trio**

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

Your testing results:

We have not found any DNA variants that could explain your medical concerns.

★ ***Here are some extra notes from your doctor about this result:***

Your Genomic Testing Results

What does this mean? There are two possible explanations for why we have not found anything:

Possibility #1: The health condition is not genetic.
Your medical concerns may not be caused by a variation in the **DNA**. Many health problems are not caused by factors related to genetics. Some examples are toxins, infectious agents, issues in pregnancy, or other harmful events. It may be very hard to pinpoint exactly what caused a person's health problems.

Possibility #2: The health condition is genetic, but we cannot detect the cause with our current knowledge. It may become possible to find a genetic cause in the future.
Genetic technology is improving, and we have a lot to learn about the **genome**. While genetic technology can read **DNA** sequences, we are still unable to interpret all of the **DNA** variants that we see. A few years from now, you may wish to request a re-assessment of your genome. This may involve a re-analysis of your sequenced genome or even a re-sequencing with newer genetic technologies.

★ 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.
You have no incidental findings.

★ Here is some space for you to take notes:

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 health conditions affecting you or your family members can be complicated and 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 tips, websites, and tools that may be useful for people who do not get a diagnosis through this testing. **Remember, you are not alone. There are many families whose genetic testing does not lead to a diagnosis.**

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.
- Your family's needs may change over time.
- The best supports focus on families' specific needs, rather than a diagnostic label.

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** and **Inclusion BC**.

Rare Disease Foundation: This foundation has a *Family Counselling Assistance Program* and a *Parent 2 Parent* Resource Network. They have a great booklet called **Living Without a Diagnosis**, which can help you plan for your future and find more resources specialized for your journey. Another similar resource is *Global Gene*'s **Toolkit for the Undiagnosed**. There is also the **Rare Disease Information and Support Line** which you can call to receive help navigating a rare disorder.

The Future Planning Tool by the **Planned Lifetime Advocacy Network** will help you build a secure future for you or anyone with a disability. It focuses on your finances, personal network, estate planning, and housing.

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.

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

Your Next Steps

Navigating your results is not something you have to do alone. It is your doctors' and/or genetic counsellors' job to make sure you understand what it means to get no genetic diagnosis from **genomic** testing, 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:

Does this mean there isn't a genetic condition? Can this result change over time? If it isn't genetic, what else can explain these health conditions? Does this result change our plans for health management? Why is there still uncertainty in genetic testing? How and when can we seek reanalysis of our **genome**?

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:** Test results may be important for the health or family planning of relatives. This includes the lack of a diagnosis. Family and friends may be better able to support you if they can inform themselves about your condition and the uncertainty of your situation. Medical testing can be a hard topic to discus with your relatives, but your healthcare team can offer guidance.
- **Your healthcare team:** It is important to keep your doctors informed about the medical testing you receive, even if no **genomic** diagnosis has been made. It is useful to share these results with your doctors because they can help you to pursue future testing or reanalysis. This may also prevent your doctors from ordering other types of tests for **genes** that have already been looked at. If you are unsure how to explain your results to your doctors, you can share this booklet with them.

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).
- Online support groups for those living with chronic, undiagnosed illness include the Undiagnosed Illness Support Group, RareConnect: Undiagnosed Diseases, and Syndromes Without a Name USA, to name a few.

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.