

Genomic Testing Results

A guide to understanding your test results and exploring your next steps

Table of Contents

(Click on the titles below to jump to that section)

Background Genomic Information	2	Health Management Tools	8
Genomic Testing You Received *	3	Financial Resources	9
Your Genomic Testing Results *	4	Support Groups	10
Incidental Findings *	5	Your Next Steps *	11
Next Steps	5		
Sharing Your Results	5		
Resources and Support	6		
Organizations and Alliances	6		

***Sections that may have been personalized for you.**

How to use this booklet

- Some parts of this booklet have been filled in by your healthcare provider. This information can be found under the orange headers marked with a star (*).
- Blue headers contain general information.
- You will see some medical terminology. These words are bolded, and are explained with in-text colour-coded definitions.
- You will see some prompts and questions that you may choose to discuss with your healthcare provider.
- If you are viewing the booklet electronically, you can click on the webpage links.

If you have any questions about your genetic testing results, please contact:

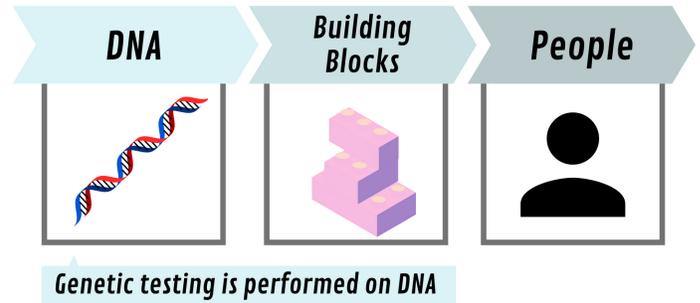


Background Genomic Information

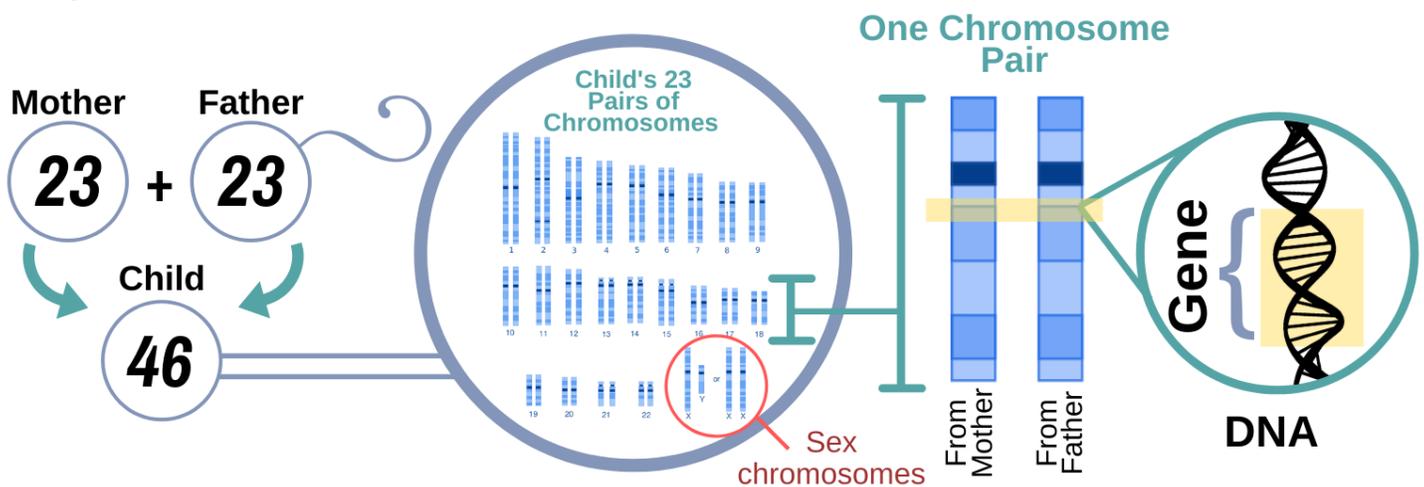
DNA is a long string of "letters."

Genes are the sections of your DNA that have instructions for making proteins.

Proteins are the building blocks of your body and play an important role in how you develop and function.



You have two copies of each **gene**, one copy from each parent. This is why you have traits that are similar to those of your parents. The interaction between these two **gene** copies determines how a certain trait will be expressed. **Your unique set of DNA, including all of your genes, is called a genome.** Your **genome** contains thousands of **genes**! Over the past few decades, scientists have been working to identify and understand these **genes**. There is still a lot of work to be done. We don't fully know the role of every part of the **genome**, which is why there can be uncertainty in genetic testing results.



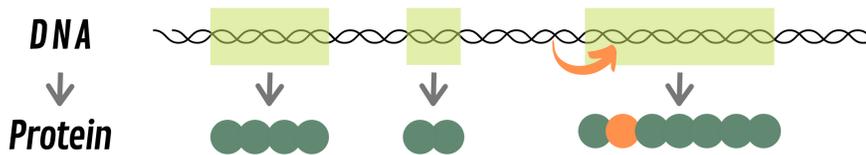
A **chromosome** is made of densely packed DNA, and carries a unique combination of genes. In this image, you can see that a child's first cell is made from 23 **chromosomes** from each parent. Because **chromosomes** come in pairs, **genes** come in pairs too. When we zoom into the cell, we see all 46 **chromosomes** (in 23 pairs). Two of our 46 **chromosomes** determine our sex. These are called sex chromosomes: males have a Y and an X chromosome (XY), while females have two X chromosomes (XX). Each chromosome will contain many **genes** that serve as instructions to produce protein.

Your Notes

Genomic Testing

Differences in the DNA, also known as **variants or mutations**, occur frequently, and most are perfectly normal. Many account for traits that are shared between family members. However, sometimes a **variant** changes the instructions in a way that alters the **protein** product. This can impact the function and development of an individual, and may cause a medical condition. Genetic technologies analyze **DNA** to identify the **variants** that may be responsible for medical concerns.

Testing You Received *



Your notes:

In the image above, the **DNA** that is highlighted in light green will be **directly involved in making protein**. This is called **protein-coding DNA**. There is still a lot of **DNA that is not highlighted in light green and does not code for protein**. This is called **non-coding DNA**. **Non-coding DNA** used to be considered "junk", however, we now know that this **DNA** can still have an impact on how proteins are made. This is shown by the orange arrow. Here, **non-coding DNA** has an impact on the protein, even if it does not directly code for it! Some tests look at all of the **DNA**, while others will only read the **protein-coding DNA**.

The genomic testing you received:

Who was tested?

Your Genomic Results

There are no genomic findings.

We completed sequencing and analysis, and have **not** found any DNA variants that could explain your medical concerns.

What does this mean?

There are two possible explanations for why we have not found anything.



Possibility #1: The medical condition is not genetic.

Your medical concerns may not be caused by a variation in the DNA. Many health problems are caused by factors that are not genetic. Other causes of medical conditions include exposure to toxins or infectious agents, lifestyle, pregnancy complications and other harmful events. It may be very hard to pinpoint exactly what caused a person's health problem.

Possibility #2: The medical condition is genetic, but we are not able to detect the genetic cause with our current technology and knowledge. It may become possible to find a genetic cause in the future, as we learn more and genetic testing technology improves.

We are working at the limits of our knowledge. Although genetic technology can read DNA sequences, we are still unable to interpret all of the DNA variation that we see. As genome science advances, new knowledge might allow us to find a disease-causing variant within your DNA sequence.

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 re-sequencing with a more advanced genetic technology. Contact the healthcare professional on the front of this booklet when you are looking to pursue these options.

Your notes:

*Incidental (Secondary) Findings **

Next Steps

Sharing Your Results

Whether you share your results and who you share them with is **entirely your decision to make**. These are some people you may choose to share the results with.

Family

Genetic information about heritable traits, including the lack of a genetic diagnosis, may be important for other family members. They may use this information to make decisions about their own health, or for family planning purposes. Furthermore, family and friends can help to support you during this time of uncertainty. Medical testing can be hard topic to discuss with your relatives, so do not hesitate to reach out to your doctor or genetic counsellor for help and guidance.

Your medical team: doctors and other allied health professionals

Despite the negative results, it is important to keep your doctors up-to-date about the medical testing you have received. Some of your doctors might already have access to your genetic testing results, while others do not. It is useful to share these results with your doctors because they can help you to pursue further genetic testing or reanalysis of your genome in the future. This may also prevent your doctors from ordering other types of testing for genes that have already been looked at. If you are uncertain of how to explain your results to your healthcare team, you can share this booklet with them.

Your notes:

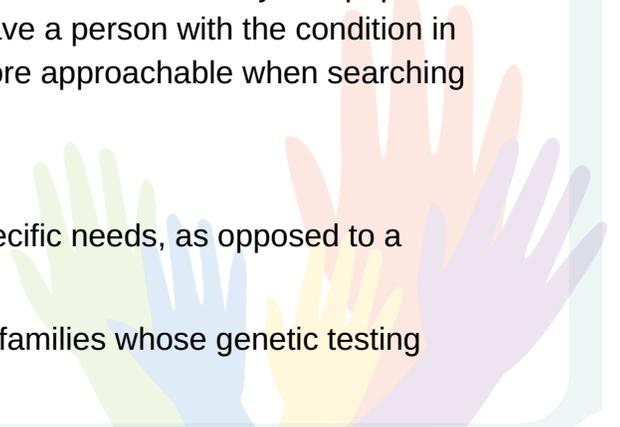
Resources and Support

Searching for resources, support and information can be a long and overwhelming process. Caring for your own health, or a family member's health, can be a complex and challenging task at times. We acknowledge that you have been putting in the hard work, and that you may already be well connected to resources that are helpful in navigating your medical concerns. Here are some additional resources that you might find helpful after receiving genetic testing results. The following pages contain general resources for healthcare management, as well as resources that are specific to those who do not receive a genetic diagnosis after testing.

Do not hesitate to reach out to your genetic counsellor or doctor if you need help navigating options and resources.

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

- Scientific literature can be very technical and complicated, and the way that papers are written may seem insensitive to readers who have a person with the condition in the family. Patient-oriented websites are usually more approachable when searching for information about your medical concerns.
- Your family's needs may change over time.
- The best support caters towards symptoms and specific needs, as opposed to a diagnostic label.
- You are not alone. There are many individuals and families whose genetic testing does not provide a diagnosis.



General Organizations and Alliances ←

In addition to these general resources, you may wish to look for organizations that target the different aspects of your medical condition. For example, if you experience seizures, the Canadian Epilepsy Alliance (<https://www.canadianepilepsyalliance.org/>) may be a helpful resource.

Family Support Institute of BC 

<https://familysupportbc.com>

Support Worker Central: This database connects families with support workers in their area. FamilyWORKs is an initiative to create employment opportunities for people with disabilities.

"The purpose of the Family Support Institute of BC is to strengthen, connect and build communities and resources with families of people with disabilities in BC."

Disability Alliance BC (DABC)

<http://disabilityalliancebc.org>

Among other services, they can provide free legal advice to those with disabilities, and they offer assistance in applying for disability benefits.

"DABC's Advocacy Access Program has been a place of support, information and one-to-one assistance for people with all disabilities."

To make an appointment, call Advocacy Access at **1-800-663-1278** (toll free).

Inclusion BC

<https://inclusionbc.org>

The Ready, Willing & Able initiative encourages employers to hire people with intellectual disabilities.

"Inclusion BC is a non-profit provincial organization that advocates for the rights and opportunities of people with intellectual disabilities and their families."

PLAN: Planned Lifetime Advocacy Network

<https://plan.ca/>

PLAN, or Plan Institute in BC, focuses on creating support networks, tools for future planning, and community initiatives.

"PLAN is non-profit organization founded in 1989 to help families secure the future for loved ones with disabilities."

Rare Disease Foundation

<https://rarediseasefoundation.org>

This foundation has a Family Counselling Assistance Program and a Parent 2 Parent Resource Network. There are meetings and events in cities across Canada, including Vancouver.

"The Rare Disease Foundation is focused on linking basic science and clinical practice to increase the efficiency of rare disease research."

This foundation has developed a resource for undiagnosed individuals. Living Without a Diagnosis:

<https://rarediseasefoundation.org/wp-content/uploads/2017/07/Living-Without-a-Diagnosis.pdf>

Contact a Family

<https://contact.org.uk>

This website contains advice and support written by parents for parents, an online parent community forum, and patient-friendly medical information on genetics and various diagnoses.

"We support families with the best possible guidance and information. We bring families together to support each other. And we help families to campaign, volunteer and fundraise to improve life for themselves and others."

This organization has developed a resource for individuals who are living without a diagnosis. You can find it here:

https://contact.org.uk/media/737817/about_diagnosis_living_without_a_diagnosis_24_july_2014_kb.pdf

Global Genes has put together a great resource to guide undiagnosed patients in becoming their own research advocate.

"Being an empowered patient means taking an equal and active role in the care of yourself or a loved one."

Becoming an Empowered Patient: a Toolkit for the Undiagnosed

https://globalgenes.org/wp-content/uploads/2014/04/GG_toolkit_six_rev3.pdf

Other Recommended Resources *

Health Management Tools ←

Curatio *Free in your app store*

Curatio is an app that connects people with similar health conditions and helps to keep track of changes in health.

"We believe everyone should have the support, information and tools they need to achieve their best health outcomes. Our mobile platform helps match patients to others that understand what they're going through, all in a private, secure environment. Curated information, daily tracking tools, evidence-based programs and fun rewards are personalized to make living with a health challenge a bit easier."

MyBooklet BC <https://mybookletbc.com>

This is a website that helps you design a personalized medical information booklet. This can help summarize medical histories and needs in a way that is easy to share with health care providers and support workers.

Medisafe Pill Reminder *Free in your app store*

This smartphone app allows you set reminders for managing your medications, and track your dosages.

HealthVault, Medical Records, myPHR, Healthspek, My Medical *Free in your app store*

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 on these apps before uploading any personal information. You may wish to de-identify all of the information before uploading.

Financial Resources and Subsidized Programs



Disability Tax Credit – <https://www.canada.ca/en/revenue-agency/services/tax/individuals/segments/tax-credits-deductions-persons-disabilities/disability-tax-credit.html>

"The disability tax credit (DTC) is a non-refundable tax credit that helps persons with disabilities or their supporting persons reduce the amount of income tax they may have to pay."

Child Disability Credit – <https://www.canada.ca/en/revenue-agency/services/child-family-benefits/child-disability-benefit.html>

"The child disability benefit is a tax-free monthly payment made to families who care for a child under age 18 with a severe and prolonged impairment in physical or mental functions."

Children and Youth with Special Needs (CYSN) Program BC

<https://www2.gov.bc.ca/gov/content/health/managing-your-health/child-behaviour-development/special-needs>

Registered Disability Savings Plan (RDSP) – <https://www.canada.ca/en/revenue-agency/services/tax/individuals/topics/registered-disability-savings-plan-rdsp.html>

"A registered disability savings plan (RDSP) is a savings plan that is intended to help parents and others save for the long term financial security of a person who is eligible for the disability tax credit (DTC)."

The government of Canada can contribute up to \$3 for every \$1 you put in.

Future Planning Tool by the Plan Institute – <https://futureplanningtool.ca/>

Build a plan to help you secure the future for you or anyone with a disability.

The Special Needs Planning Group – <http://www.specialneedsplanning.ca/index.html>

"This Web site is presented to you as a resource which will provide you with some basic information necessary to the understanding and implementation of plans for the future of your family member with a disability."

This parent-made website highlights legal and financial considerations for long term planning.

The At Home Program

<https://www2.gov.bc.ca/gov/content/health/managing-your-health/child-behaviour-development/special-needs/complex-health-needs/at-home-program>

"This program is intended to assist parents or guardians with some of the extraordinary costs of caring for a child with severe disabilities at home."

Nursing Support Services

<http://www.bcchildrens.ca/our-services/sunny-hill-health-centre/our-services/nursing-support>

"We are community-based registered nurses throughout BC who assist children and youth with medical complexities to live in their homes and in their communities."

Travel Assistance Program (for non-emergency medical services)

<https://www2.gov.bc.ca/gov/content/health/accessing-health-care/tap-bc/travel-assistance-program-tap-bc>

Finding a Support Group



Individuals undergoing similar experiences can connect through support groups. These groups create a space for sharing experiences and feelings, building community, and learning from others. They may provide both emotional support and firsthand information. Support groups come in many different structures: some are face-to-face while others are found on online platforms, and they can be either private or open to the general public. Listed below are some platforms you may use in finding support groups. **If you do not have a diagnosis, you can also search for groups that are based on some of the symptoms you share.** Make sure to check the privacy and security conditions for each platform before sharing your information online.

- **Facebook** fosters many local and international support groups. In the Facebook search bar, type [Disease Name] followed by [Location]. For example, "Cerebral Palsy Vancouver."
 - Some of these groups may be closed to the public in order to create a more private setting for the disease community. In this case, you will have to request to join by clicking "join group".
 - **You can also find more general support groups that are still helpful, supportive and informative. Try searching for "rare disease" or "complex kids" groups.**
- **Canadian Directory of National Support Groups**
<https://www.lhsc.on.ca/canadian-directory-of-genetic-support-groups/introduction-to-the-directory>
This directory will take you to a webpage specific to your health condition.
- **RareShare** <https://rareshare.org/>
Create an account to join closed support groups specific to rare genetic conditions.
 - Here is a guide on how to get started: <https://rareshare.org/howtos/welcome-to-rareshare>
 - RareShare also has podcasts on specific conditions, as well as general podcasts on living with rare diseases.

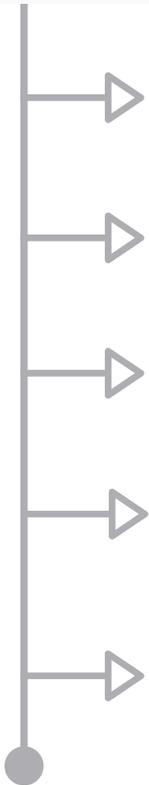
Online support groups for those living with a chronic, undiagnosed illness:

- **Undiagnosed Illness Support Group:**
<https://www.facebook.com/groups/588261638030294/>
- **Rare Connect: Undiagnosed Diseases:**
<https://www.rareconnect.org/en/community/undiagnosed-diseases>
- **SWAN USA (Syndromes Without a Name):** <https://www.facebook.com/swanusa/>
- **RareShare: Undiagnosed:** <https://rareshare.org/communities/undiagnosed>
- **Self Management BC:** <https://www.selfmanagementbc.ca/default.aspx?lang=1&seo=communityprograms&>

Extra notes from your healthcare provider *

Your Next Steps

Along with genetic testing results comes a lot of information to process and consider. Take your time. Here is some space to keep track of actions you would like to pursue. This may include connecting with a certain resource, visiting your family doctor, searching for a specialist, or any other changes in health management. If you need support with this process, or still have questions about your results, **do not hesitate to contact your genetic counsellor or doctor.** Contacts are listed on the front page of this booklet.



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 Canadian Association of Genetic Counsellors Find a Clinic tool:** <https://www.cagc-accg.ca/?page=225>
- **The National Society of Genetic Counselors Find a Genetic Counselor tool:** <http://nsgc.org/p/cm/ld/fid=164>