LOCK

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)

★ 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 teal headers marked with a star (★) and in blue text boxes.
- You will see some medical terms. These words are bolded, and are explained with in-text colour-coded definitions.
- 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:

The Genomic Results Booklet was created by Julia Handra, Isabel Jordan, Brenda Lenahan, Kelsey Ohs, Colleen Guimond, Patricia Birch, and the Friedman Lab, with the support of the CHILD-BRIGHT Network.

Version 01-06-2021

Results Summary ★

Date: today

Name:

Click on the page number to jump to that section.

Genomic testing received: Multigene Panel

Page 4

Multigene panels read the genetic instructions, also known as DNA, in a set of genes that are linked to certain health conditions. This panel looks for differences in the spelling of the DNA in just this set of genes. These differences are known as variants.

Who received testing? Singleton: Just the person with the health condition.

Genomic testing result:

There are no findings that explain your health concerns.

What does this mean?

Page 5

There are two possible explanations for why we have not found a genetic cause:

1. Your health condition is not genetic. There is no genetic cause.
2. There may be a genetic cause, but we are unable to detect it with our current technology and knowledge

Incidental (secondary) findings

Page 6

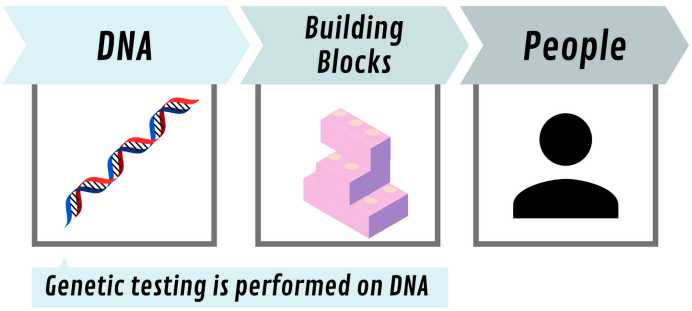
Before testing, you stated that you did not want to find out about incidental findings.

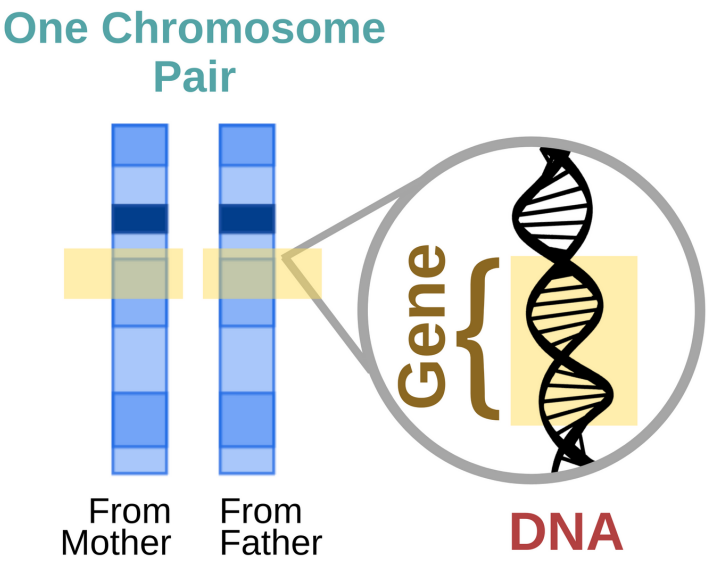
Notes from your healthcare provider

The rest of the booklet explains this information in more detail, and offers some resources.

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.





Genes are packaged in groups, called **chromosomes.** You have 23 **chromosomes** from your mother, and another matching 23 from your father.

Because **chromosomes** come in pairs, **genes** come in pairs too. 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 show up.

Your unique set of DNA**,** including all of your genes, is called a **genome**. Your **genome** contains thousands of **genes**!

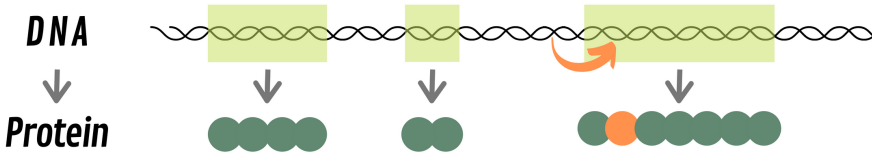
Over the past few decades, scientists have been working to identify and understand these **genes**. There is still a lot of work to be done. We don't fully know the role of every part of the **genome**, which is why there can be uncertainty in genetic testing results.

Your Notes

Genomic Testing

Differences in the DNA are known as **variants or mutations.** They 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 may cause a medical condition. Genetic technologies analyze **DNA** to find the **variants** that may cause medical concerns.

Testing You Received ★



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 in between. This is called **non-coding DNA**. **Non-coding DNA** can still have an impact on how proteins are made. For example, it may accelerate or slow down the creation of proteins.

Some tests look at all of the **DNA**, while others will only read the **protein-coding DNA**.

Your notes:

The genomic testing you received: Multigene Panel

Multigene panels read the genetic instructions, or DNA, within a limited number of genes. Scientists have developed lists of genes likely to cause certain medical conditions. Differences in the spelling of the DNA within these genes may explain your health concerns. Multigene panels only read the DNA that directly codes for protein (protein-coding DNA) within the genes on the list. Panels cannot detect variants in other genes not on the list. A multigene panel might only look at one of two of the DNA sections highlighted in green on the diagram above.

Who was tested?

Singleton: Just the person with the health condition.

Your Questions

Navigating genomic results is not something you have to do alone. Your doctors and/or genetic counsellors are there to support you through this process. It is our job to make sure that you understand what the genomic results mean for your family and support you through your next steps. We are best able to do so when we know more about your needs. You can help us understand these needs by asking us for the information that is most important to you.

Here are some questions patients often ask their doctor or genetic counsellor, and some space for you to keep track of your own questions.

Were you able to find a change in DNA that explains the symptoms?

Yes / Possibly

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

No

- Does these mean there isn't a genetic condition? Can this result change over time?
- If it isn't genetic, what else can explain these health concerns?
- What does this mean for our health management?

Your Genomic Results

There are no genomic findings.
We 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. Some examples are toxins, infectious agents, lifestyle, issues in pregnancy and other harmful events. It may be very hard to pinpoint exactly what caused a person's health problem.

Possibility #2: The health condition is genetic, but we cannot detect the cause with our current technology and knowledge. Genetic technology is improving and we are still learning. It may become possible to find a genetic cause in the future.

We are working at the limits of our knowledge. While 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 genetic cause in your DNA.

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 newer genetic technology. Contact your doctors when you are looking to pursue these options.

Your notes:

Incidental (Secondary) Findings ★

Before testing, you stated that you did not want to find out about incidental findings.

While searching through your genome for variants that may be causing your health condition, we can seek and report additional health information. Such information is called an incidental (secondary) finding. Incidental findings are not related to the reason you received genetic testing. You decided that you did not want us to seek or report incidental findings.

Next Steps

Sharing Your Results

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

Family
Genetic information about heritable traits may be important for other family members. This includes the lack of a diagnosis. They may use this information to make decisions about their own health or for family planning. Family and friends can also help to support you during this time of uncertainty. Medical testing can be hard topic to discuss with your relatives. Your doctor or genetic counsellor can offer guidance.

Your medical team: doctors and other health professionals
It is important to keep your doctors informed about the medical testing you receive. This is true even if there are no findings. 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 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 uncertain of how to explain your results to your healthcare team, you can share this booklet with them.

Your notes:

Resources and Support

Caring for your own health, or a family member's health, can be complex and challenging at times. While you just received genetic testing, we know that you have been putting in the hard work long before. You may already be well connected to resources that help you manage your medical concerns. Here are some other tips, websites and tools that may be helpful after receiving genetic testing results.

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...

- Science literature can be very technical and hard to read. The way that papers are written may seem insensitive to patient-readers, or their family members. Patient websites are often more friendly when looking for information on a diagnosis.
- 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

Along with these general links, you may wish to look for resources 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://cdn.shopify.com/s/files/1/0267/4688/0097/files/Living-Without-a-Diagnosis.pdf?v=1587664879

☐ 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/wp-content/uploads/2021/03/living_without_a_diagnosis.pdf

Global Genes

globalgenes.org/

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

People who have similar experiences can connect through support groups. These groups create a space for sharing experiences and feelings, building community, and learning from others. They can provide both emotional support and firsthand information. Support groups come in many different forms. Some are face-to-face while others are found on online platforms. 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.**

- **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/articles/how-to-use-the-rareshare-network-for-rare-disease-patients
    - RareShare also has podcasts on specific conditions, as well as general podcasts on living with rare diseases.

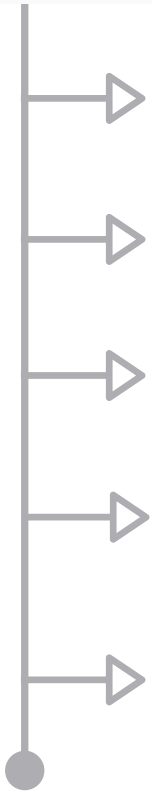
Online support groups for those living with a chronic, undiagnosed illness:

- **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/CommunityResources

Extra notes from your healthcare provider ★

Your Next Steps

There is 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, please contact your genetic counsellor or doctor.**



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:** *https://findageneticcounselor.nsgc.org/*