LOCK

Genomic Testing Results

A guide to understanding genomic test results and exploring next steps

Table of Contents

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

★ Some parts of this booklet have been filled in by a healthcare provider. This information can be found under the headers marked with a star.

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

The Genomic Results Booklet was created by members of the Friedman lab and parent partners, with the support of the CHILD-BRIGHT Network.

Version 20-10-2021

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 test looks for differences in the spelling of the DNA in only this set of genes. These differences are known as variants.

Who received testing? Trio: the person with the health condition and family members.

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 child's 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 5

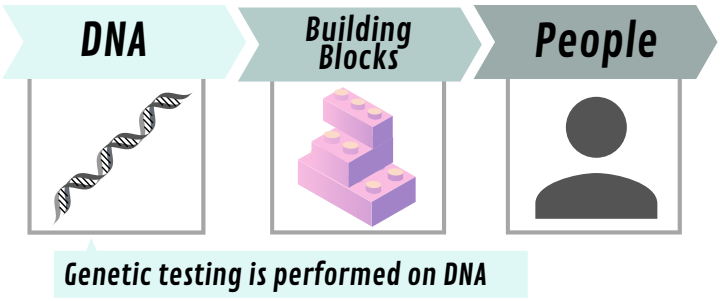
There was an incidental finding. Please refer to your child's doctor for more information.

Notes from your child's healthcare provider

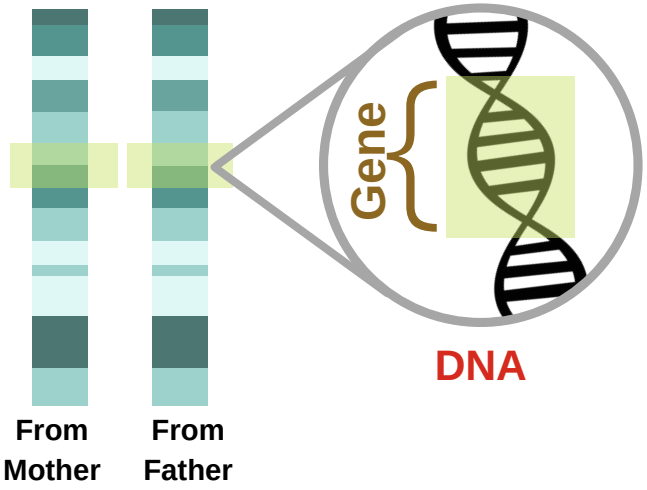
The rest of the booklet explains this information in more detail, and offers some resources.

Background Genomic Information

DNA is a long string of "letters." **Genes** are sections of DNA that have instructions for making proteins. **Proteins** are the building blocks of the body and play an important role in development and function.



One Chromosome Pair



Genes are packaged in groups, called **chromosomes.** Each person has 23 **chromosomes** from their mother, and another matching 23 from their father. Because **chromosomes** come in pairs, **genes** come in pairs too. We have two copies of each **gene**, one copy from each parent. This is why children have traits that are similar to those of their parents. The interaction between these two **gene** copies determines how a certain trait will show up.

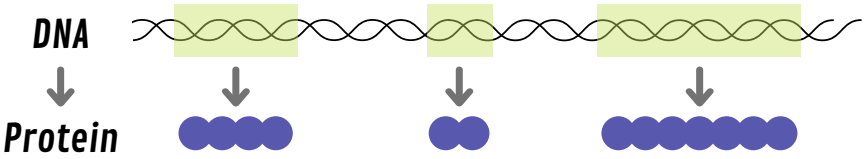
Everyone's unique set of DNA**,** including all of their genes, is called a **genome**. The **genome** contains thousands of **genes**!

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 Recieved ★

Differences in the DNA are known as **variants or mutations.** They occur frequently, and most are perfectly normal. Many **variants** 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.



In the image above, the **DNA** that is in green boxes 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 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 child's 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 or two of the DNA sections in green boxes on the diagram above.

Who was tested?

Trio: the person with the health condition and family members.

Your Genomic Results

There are no genomic findings

We have **not** found any DNA variants that could explain your child's 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 child's 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 child's DNA.

A few years from now, you may wish to request a re-assessment of your child's genome. This may involve a re-analysis of your child's sequenced genome or even re-sequencing with newer genetic technology. Contact your child's doctors when you are looking to pursue these options.

Incidental (Secondary) Findings ★

There was an incidental finding. Please refer to your child's doctor for more information.

While searching through the genome for variants that may be causing a patient's health condition, the lab may also report additional health-related DNA variants. These are called incidental (secondary) findings because they are not related to the reason for receiving genetic testing, but can be important nonetheless. You decided that you would want to be informed about incidental findings. Your child's doctor will give you more information about the incidental findings.

Next Steps

Your Questions

Navigating these results is not something that needs to be done alone. Healthcare providers are there to provide support through this process. It is their job to make sure you understand what the results mean for your family and help you through next steps. They are best able to help when they know about your family's needs. You can help them understand these needs by asking for the information that is most important to you. Here are a few questions that some families ask their healthcare providers, and some space for you to keep track of your own.

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

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?

Sharing Your Results

Whether you share your family's 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.

Your medical team: doctors and other health professionals
It is important to keep your child's doctors informed about the medical testing they receive. This is true even if there are no findings. Some doctors might already have access to your child's genetic testing results, while others do not. It is useful to share these results with your child's doctors because they can help you to pursue future testing or reanalysis. This may also prevent the doctors from ordering other types of tests for genes that have already been looked at.

Resources and Support

Caring for your own health, or a family member's health, can be complex and challenging at times. You may already be well connected to resources that help you manage your child's medical concerns. Here are some other tips, websites and tools that can be helpful after receiving genetic testing results. These pages contain both general and case-specific resources.

Do not hesitate to reach out to your child's doctor or genetic counsellor if you need help in 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, rather than 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

☐ **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."

Living Without a Diagnosis (a pamphlet):
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."

This organization has developed a resource for individuals who are living without a diagnosis:
https://contact.org.uk/wp-content/uploads/2021/03/living_without_a_diagnosis.pdf

☐ Global Genes *https://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

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*
"An 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 website 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*

Health Management Tools

Curatio *https://www.curatio.me/*
Connect to people with similar health conditions and keep track of health changes.

HealthVault, Medical Records, myPHR, Healthspek, My Medical *Free in your app store*
Track medical history, store results from medical tests, and share medical information.

MyBooklet BC *https://mybookletbc.com*
Design a personalized medical information booklet to share with healthcare providers.

Finding a Community

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. Some are face-to-face while others are found on online platforms. 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 - Undiagnosed group:** https://rareshare.org/communities/undiagnosed
    - 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:

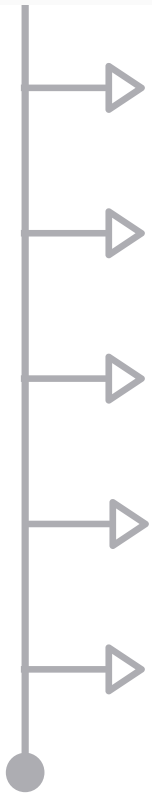
- **Undiagnosed Illness Support Group:** *https://www.facebook.com/groups/588261638030294/*
- **SWAN USA (Syndromes Without a Name):** *https://www.facebook.com/swanusa/*
- **Self Management BC:** *https://www.selfmanagementbc.ca/CommunityResources*

Other Recommended Resources ★

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 child's 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/*