

Understanding Genetic Testing

Table of Contents

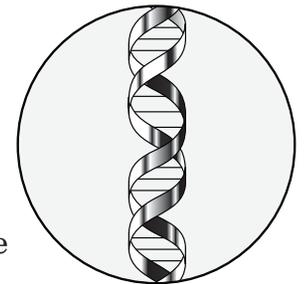
Introduction	3
What is a Gene?	3
What are Chromosomes?	4
How Do Genes Cause Disease?	4
How Do People Get Genetic Disorders?	4
How are Genetic Conditions Inherited?	5
Patterns of Inheritance	6
How Do Genes Mutate?	11
What is Genetic Testing?	14
Is Genetic Testing for Me?	14
How is Genetic Testing Done?	15
What are the Types of Genetic Testing?	16
What is Genetic Testing Used For?	17
What are the Benefits to Genetic Testing?	18
What are the Limitations of Genetic Testing?	18
What are the Disadvantages of Genetic Testing?	20
What do the Results Mean?	21
How Genetic Testing is Funded?	24
What Will Happen to My Sample After the Test?	24
Providing Consent	25
Who Can Help Me Learn More About Genetic Testing?	25
Definitions	26
References	30

Introduction

The purpose of this booklet is to provide you and your family general information about genetics and genetic testing. Understanding how our genes cause disease can be challenging. This booklet aims to answer your basic questions, explain how your genes may cause disease and how genetic disorders may be inherited. It also provides information about the testing process and what you should know before you decide to complete genetic testing. Some sections of this booklet may or may not apply to you or your family. Your physician, clinic nurse, or a genetic counsellor will discuss how parts of this booklet apply to you. All genetic tests and the results of genetic testing will be discussed with you at your clinic appointment.

What is a Gene?

Genes are found in all of the cells, tissues, and organs of our bodies. Genes contain many instructions for specific activities that help our bodies grow and function. Genes also have the information that make you look the way you are.

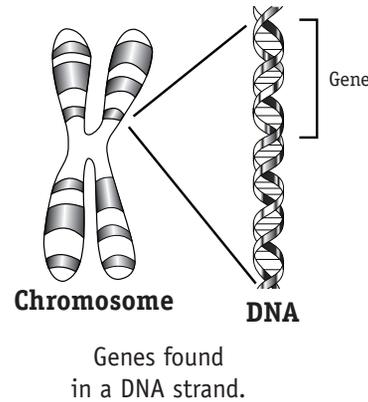


We all have slight differences in our genes that make us special. For example, the genes in our bodies determine the colour of our eyes and hair, how tall we are, and how our bodies digest sugar. These traits that are determined by our genes are passed from parent to child. This is called inheritance.

Genes are made of a very small string-like substance called **DNA**, which contains four different chemicals like four letters of the alphabet. These letters can spell out all of the genes in our body.

What are Chromosomes?

All of our genetic material (DNA and genes) are tightly held together by **chromosomes**. Chromosomes are found in all cells of the body. Human cells normally have 22 pairs of chromosomes and 1 pair of sex determining chromosomes. Half of each pair is inherited from the mother and the other half from the father.



How do Genes Cause Disease?

Most of our genes work to help our bodies grow and function as a healthy person. However, when there is an abnormal change in a gene, it may cause a disease or increase a person's risk of developing a **genetic disorder** or **genetic disease**. Abnormal changes in genes, or **mutations**, happen due to an altered spelling of our DNA and could cause the genes to no longer work as they should in our bodies.

How Do People Get Genetic Disorders?

You receive half of your genes from your mother and half from your father. You pass half of your genes to your children. Genes are passed from parent to child. This is called **inheritance**. It is not possible to control which genes are passed, therefore inheritance is all by *chance*.

This could mean that there is a *chance* a mutated gene was inherited. However, there is also a *chance not* everyone will get the mutated gene even if your parent has it.

It is also important to know that not all mutations to our genes are passed from our parents. Sometimes, **people can get genetic disorders if they have copying mistakes or damages in their own DNA**.

People who have abnormal changes in their genes but do not have the disease are called **carriers**. Carriers also have a chance in passing their changed gene to their children. This chance depends on what gene is changed and whether *one* or *both* your parents have the changed gene.

How are Genetic Conditions Inherited?

Genetic conditions caused by a mutation to a single gene can be inherited by one of several patterns. The patterns of inheritance include **autosomal dominant**, **autosomal recessive**, **X-linked recessive**, and **X-linked dominant**. The pattern of inheritance depends on what gene is mutated and where the mutated gene is located.

Key Words to Know...

It is important to know and understand the following terms.

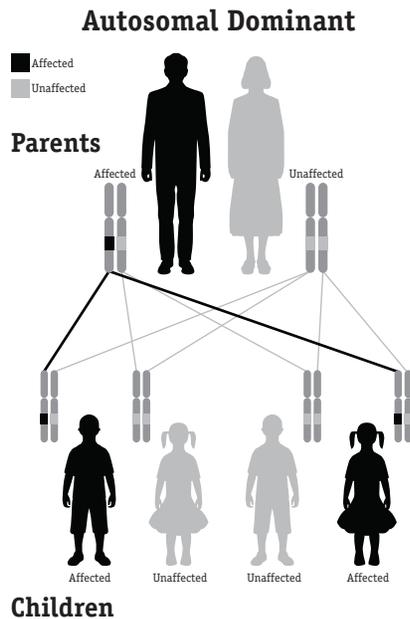
- **Autosomal** - The mutated gene that is responsible for the genetic disorder is located on one of the 22 pairs of **autosomes**, which are non-sex determining chromosomes.
- **X-linked** - The mutated gene that is responsible for the genetic condition is located on the X-chromosome (one of the sex determining chromosomes).
- **Dominant** - A genetic condition that only requires one copy of the mutated gene from one parent.
- **Recessive** - Genetic conditions that require two copies (one copy from both parents) of the mutated gene.

Patterns of Inheritance

Autosomal Dominance

Autosomal Dominant conditions are expressed in people who have just one copy of the mutant gene. Affected males and females have an equal chance of passing on the trait to their children. Affected people have one normal copy of the gene and one mutant copy of the gene, thus each child of an affected parent has a 50% chance of inheriting the mutant gene.

One mutated copy of the gene in each cell is enough for a person to be affected by an autosomal dominant disorder.

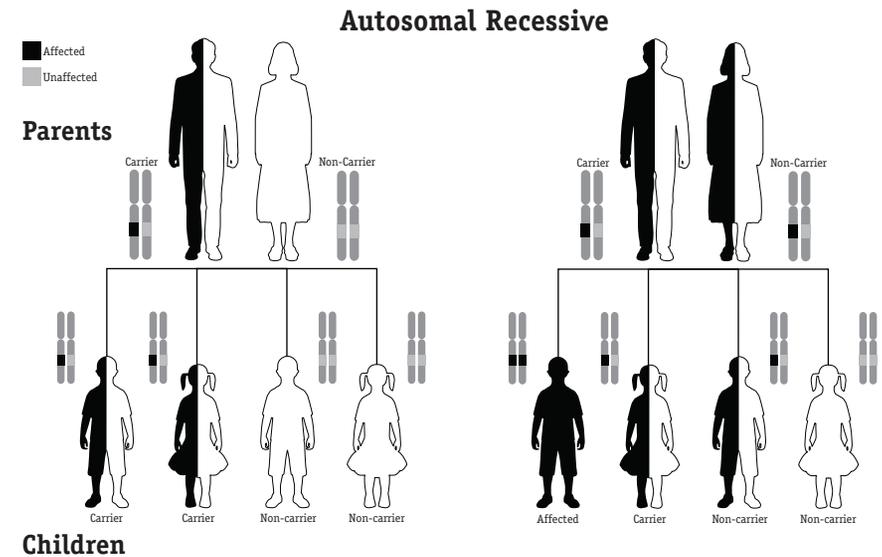


In this example, a man with an autosomal dominant disorder has two affected children and two unaffected children.

Autosomal Recessive

Autosomal recessive conditions can show signs of the disease only when the person has two copies (one copy from each parent) of the mutated gene. If the person only has one copy of the mutated gene but does not develop the condition, the person is a **carrier**. Females and males are affected equally by traits from the autosomal recessive inheritance.

If both parents are carriers, there is a 25% chance of their child being unaffected; a 25% chance of being affected; or a 50% chance of being an unaffected carrier. Autosomal recessive disorders are typically not seen in every generation of an affected family.



This shows the chances an offspring who could develop a genetic disease if one parent or both parents carry a genetic disorder.

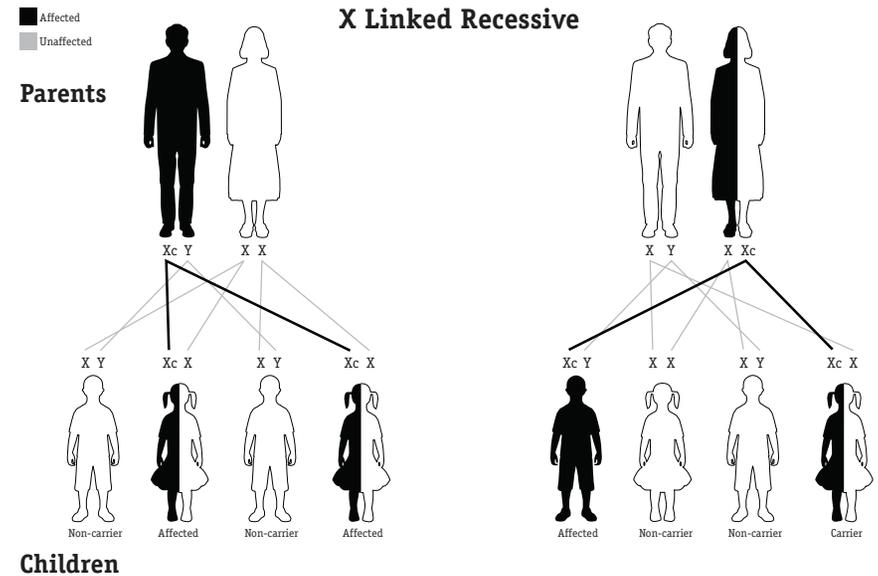
X-linked Recessive

X-linked recessive conditions are also caused by mutations in the genes on the **X-chromosomes**. These conditions do not show any signs and symptoms of the disease if the person has one normal copy and one mutated copy of the gene.

However, males who have the X-linked recessive mutated gene will fully display the genetic condition because they only have one copy of the X-chromosome. This is because males only have one X-chromosome whereas females have two X-chromosomes. Males do not have a normal copy of the X-chromosome to balance out the mutated copy.

For that same reason, women are rarely affected by X-linked recessive diseases. Females only fully display the genetic condition if they have two copies of the mutated gene.

There is no father to son transmission since the X-chromosome will come from the mother. However, there is father to daughter and mother to daughter and son transmission. If a man is affected with an X-linked recessive condition, all his daughters will inherit one copy of the mutant gene from him.



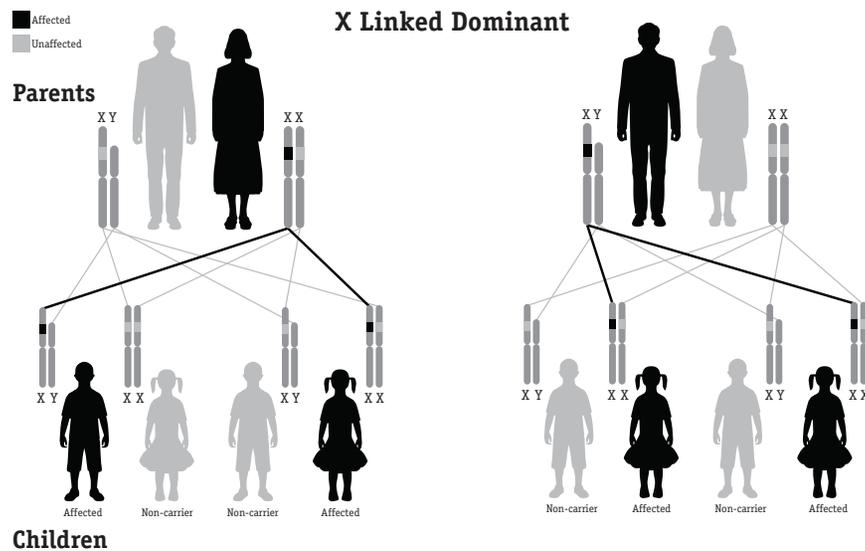
The father is affected and the mother has the normal gene. The affected father has a 100% chance in passing his mutated gene to all his daughters

The mother is a carrier and the father has the normal gene. A carrier mother has a 50% chance in passing her mutated gene to her children. In this case, the baby boy is fully affected and the baby girl will become a carrier.

X-linked Dominance

A person with an **X-linked dominant** gene will only need one copy of the mutant gene to have signs and symptoms of the genetic disease. X-linked dominant disorders are caused by mutations in genes on the X chromosome.

Since the gene is located on the X chromosome, a father will not transmit the mutated gene to his son; however, the mutated gene can be passed on to his daughter. This happens because the father has only one X chromosome to give so all his daughters will be affected. An affected mother can also pass on the mutated gene to her child, but there is only a 50% chance her child will inherit the X chromosome.



A father with a X-linked dominant condition will not pass the affected gene to his sons but will pass it to all his daughters.

A mother with a X-linked dominant condition has a 50% chance of passing the affected gene to all her children.

How Do Genes Mutate?

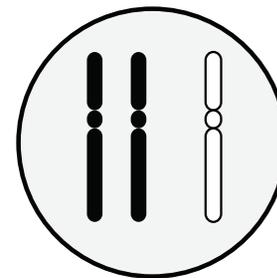
It is important to know that not all mutations to our genes are passed from our parents. **People can get genetic disorders if there is an error in their own genes.**

Errors that cause mutations can happen in several ways. It can happen during cell division when the cells in our body try to make copies of each other. In every cell's centre, there is a **nucleus**. This nucleus is the control centre of a cell and it is where genetic material such as your DNA and chromosomes are found. Any changes to the chromosome number or structure can cause chromosome abnormality which can lead to different genetic conditions and diseases. Types of chromosome abnormality are **Numerical Abnormalities** and **Structural Abnormalities.**

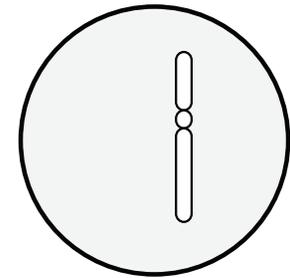
Numerical Abnormalities

Numerical abnormalities happen when a person is missing a chromosome from a specific pair called **monosomy** or a person has more than two chromosomes of a specific pair called **trisomy**.

For example, Down Syndrome, also known as Trisomy 21, is caused by a numerical abnormality. A person with Down Syndrome has 3 copies of chromosome 21 instead of having the normal number of 2 copies.



Trisomy



Monosomy

Structural Abnormalities

Structural abnormalities can happen in several different ways. It can be caused by any deletion, duplication, translocation, and/or inversion to the chromosomes.

Deletion is when a section of the chromosome is missing or deleted.

Duplication is when there is an extra copy in a section of the chromosome.

Translocation is when a section of one chromosome is transferred to another chromosome. There are 2 types of translocations, Reciprocal translocation and Robertsonian translocation.

Reciprocal translocation happens when sections from 2 different chromosomes have been exchanged

Robertsonian translocation happens when an entire chromosome attached itself to another chromosome

Inversion happens when a section of the chromosome breaks off and turns upside down before it is reattached. The genetic material is then inverted.

Ring happens when a section of the chromosome breaks off and forms into a ring. It can happen with or without losing any genetic material.

Maternal Inheritance

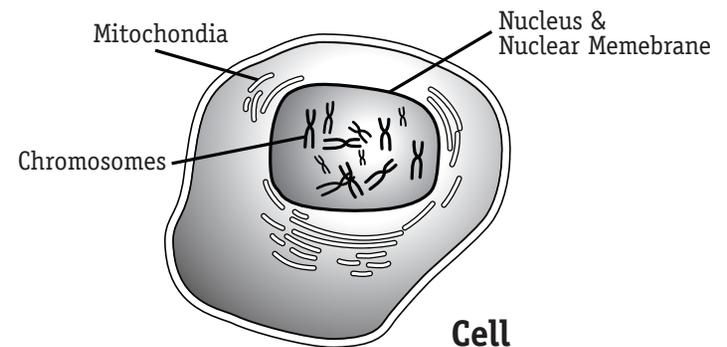
Maternal inheritance is a type of inheritance which involves passing down genes in the mitochondrial DNA from the mother to the child. **Mitochondria** are tiny, cucumber-shaped “factories” that turn the food we eat into energy that are used in all body cells. It is important to understand that mitochondria found in our cells also contain DNA that could be passed from parent to child.

Disorders caused by mitochondrial DNA mutations can appear in every generation of a family and can affect both males and females. However, mitochondrial DNA is only inherited from the mother and only her daughters can pass it on to the next generation.

Mitochondrial DNA

Homoplasmy is a term used in genetics to describe a cell whose copies of mtDNA are all identical. However, people with a mutation to their mitochondrial DNA can have **heteroplasmic cells** where each cell in the body can have a mixture of good and bad mitochondrial cells.

The severity of the mitochondrial condition can be determined by the amount of mutated (changed) mitochondrial cells in the cells of the body. For example, if a cell contains 70% mutated mitochondria, then the person will likely display more symptoms than a person with a cell that only contains 30% of the mutated mitochondria.



This is a cell with nuclear and mitochondrial DNA

What is Genetic Testing?

Genetic testing is a group of tests used to find changes in the human genes. The results of a genetic test, along with other assessments, are used as a piece of evidence to either confirm or rule out a certain genetic disorder. It can also be used to determine if there is a *chance* for an affected person to pass on the genetic condition to his or her child.

Is Genetic Testing for Me?

Genetic testing can sometimes:

- help diagnose a disease
- find out if a person is a carrier for a genetic disease
- find genetic diseases in a developing baby during pregnancy
- show whether a person has inherited a certain disease before symptoms start
- help doctors choose correct treatment and management for their symptoms

Results of genetic testing may not always provide the above information. It will not detect the severity of the disease. Sometimes, it will not give us a definite “yes” or “no” answer. It is also important to understand that not all genetic diseases have treatments.

How is Genetic Testing Done?

Prior to genetic testing, a health care provider will need to collect a small sample of tissue from your body. The types of tissues collected for genetic testing may include:

- blood
- urine
- cells swabbed from inside your mouth
- saliva
- hair
- skin
- tumours
- muscle tissue
- amniotic fluid that surrounds a fetus during pregnancy

After the sample is collected by your health care provider or local laboratory, the sample will be sent to a specialized laboratory for testing.

What are the Types of Genetic Testing?

There are many types of genetic tests available. Different types of genetic testing can be used for different reasons. It is important to discuss with your physician which tests are suitable for you. Some common types of genetic tests are:

Gene test

This test looks at the person's genetic make-up and finds gene changes that may or may not cause a genetic disease.

Chromosomal Tests

This test looks at a person's chromosomes and can find changes to the chromosomes. Human cells normally have 23 pairs of chromosomes. Half of each pair is inherited from the mother and the other half from the father. For example, chromosomes can be deleted, switched, or expanded which may cause a genetic disorder.

Biochemical Tests

This test can measure the amount of a **protein** or the activity of an **enzyme** responsible for the makeup or breakdown of tissues in the body. For example, it can detect metabolic conditions responsible for the breakdown of food which provide energy to the body. In this case, the test result could help determine which foods to avoid that may increase symptoms from the metabolic disease. It is important to understand that not all genetic or metabolic diseases have treatments.

What is Genetic Testing used for?

Diagnostic Testing

This test is used to find or rule out a genetic disorder. It may also help to find the gene changes that are causing the signs or symptoms. It is important to know that this test may not detect all genes or all genetic conditions.

Predictive or Pre-symptomatic Testing

This test is used for people who do not have signs or symptoms but have a family member that has a genetic disorder. This test can find out if you have the gene that causes the disease. It can also find out if the changes in your genes may increase your risk of getting a genetic disorder later in life. For example, if your mother is diagnosed with breast cancer, you may want to determine whether you have the gene that may cause breast cancer later in life.

Newborn Screening

This test is used to look for rare disorders. It is done by pricking a needle at the heel of the foot to get a blood sample. It can also help find out if your baby has a genetic disorder that can be treated and managed early in life. If a screening test suggests a problem, your baby's doctor may do more testing.

Prenatal Testing

Certain genetic tests may be offered to pregnant women to find gene changes in the unborn baby. It can help parents make decisions about the pregnancy and can lower their uncertainties. However, prenatal testing cannot find all possible inherited disorders or birth defects. Prenatal testing only looks for the most common inherited disorders.

What are the Benefits of Genetic Testing?

Knowing the results of the test can help a person:

- feel less uncertain
- feel less worried about getting a disease
- find out if a disease is developing
- find out whether a disease can be prevented or treated
- make informed choices about his or her future, such as having a baby, making a career choice, and choosing insurance coverage
- find out if lifestyle changes will reduce the risk of getting a disease
- get proper treatment and management for the symptoms

What are the Limitations of Genetic Testing?

Genetic Test Results do not give all the answers

A gene change or mutation can happen anywhere in our genetic make-up. Sometimes, a single gene may have more than one type of mutation. Depending on the mutations, a person may show more or less symptoms of a disease. Some mutations may not cause symptoms at all. And in some situations, more than one mutation must be present in order for a person to have the disease.

Results of genetic testing cannot confirm if a person will for sure have a disease, how severe the condition will be, or when symptoms will begin. **This means that the result of the test may not provide all the answers.** Genetic testing only provides limited information on a genetic condition.

Labs cannot detect all Gene Changes

It is also important to understand that most laboratories can only test for the most common gene changes. It is possible that a person could carry a rare gene change that may not have been detected during the test. The genetic technology today may not be advanced enough to detect all gene changes.

Medical and Family History are Important in Genetic Testing

Before undergoing a genetic test, it is very important to provide accurate information about your medical and family history to your health care provider. Knowing this information can help health care providers decide which test is needed to determine your condition.

Errors in Sample Handling

All certified laboratories have strict rules for handling samples that are sent to them. In rare cases, problems may occur in handling the sample, which may cause incorrect results. Some examples include wrong labelling, contamination, or misinterpretation of the laboratory findings. If this happens, the laboratory may contact your health care provider and require you to give a second sample.

Genetic Testing cannot always give a “Yes” or “No” Answer

Genetic testing continues to grow as new technology to find gene changes improves. Genetic testing can find changes in our genes but sometimes the lab does not know if the changes are the cause of your symptoms and further testing may be required. The results in this case are said to be **equivocal**. Therefore, **genetic testing cannot always give us a simple “yes” or “no” answer.**

What are the Disadvantages of Genetic Testing?

Emotional Burden

A big concern that many people have with genetic testing is how the result may affect a person's future. The decision to have genetic testing can be very stressful. **The result of a genetic test can take many months to return from the laboratory.** During this time, you may experience emotions of worry, confusion, and uncertainty. For these reasons, it may be best to meet with your health care provider to discuss any questions or concerns.

Exposure of Family History

Sometimes a positive test may reveal information not only about you but about your relatives. A positive result may change family relationships as the result may disclose information about the genetic make-up of other relatives, even if they have not been tested. Furthermore, a genetic test may reveal unexpected relationships, such as having a non-biological relative. You may have to make a decision to discuss your condition with your family members if they are also at risk for the genetic disease.

Insurance Coverage

In Canada, primary health insurance is not affected, but there can be problems in getting life insurance, extended or private health insurance, private disability insurance or mortgage insurance if you are diagnosed with a disorder. If you do not have any signs or symptoms of a genetic disorder, you may want to look into insurance coverage before being tested.

What Do the Results Mean?

Genetic test results are not always straightforward to understand. It will not always give a simple “yes” or “no” answer. The results are often hard to interpret and explain. It is important to understand the meaning of results by asking questions before and after the test.

Positive Results or “Abnormal”

A positive or “abnormal” test result means you have a gene change that can cause a specific condition or can put you at a higher risk of developing the disease in the future. However, not all positive test results mean you will definitely get the disease. Furthermore, not all gene changes lead to symptoms of a genetic disease. This will depend on the type and number of gene changes found.

Negative Results or “Normal”

A negative test or “normal” result is usually good news, however, a negative test result may not rule out a genetic disease that is related to a gene change. For example, you may still have the gene change, even if your test result is “normal”. For this reason, some negative results may be inconclusive. It is possible that the test may have missed the disease causing gene. As a result, further genetic testing may be needed as there is no single genetic test that can test for all genes.

Inconclusive or Equivocal Results

An inconclusive result usually means that a gene change has not been previously identified as the cause of a specific genetic disorder.

Reasons why results come back inconclusive are:

1. the laboratory was unable to examine every section of the genes
2. the laboratory found a change in one or more of the genes, but cannot predict whether it is causing the symptoms

If possibility number 1 is the reason for your inconclusive result, your health care provider may arrange for additional genetic testing.

If number 2 is the most likely reason for this inconclusive result, then there is a change to the gene that has not been seen before. This gene change may or may not cause symptoms.

It is difficult to tell if the changed gene is a natural or disease causing change. The result does not tell us if the changed gene found will affect your health. An inconclusive result cannot confirm or rule out a certain diagnosis. It also does not tell us if a person will develop a disorder later in life. Inconclusive or Equivocal results may require additional testing of you or other members of your family. Testing additional family members may help your physician determine if the gene change is related to your symptoms.

Testing Family Members

Inconclusive or Equivocal results may require additional testing of you or other members of your family. Testing additional family members may help your physician determine if the gene change is related to your symptoms. Family members may be asked to provide a sample to assist in identifying what the gene change may mean. In some cases, this testing will help to determine how you have inherited this gene change. This testing will not show if your family member has a genetic disorder. The results of family member testing are to help identify the cause of your symptoms.

If additional family members have symptoms similar to your own a referral to the Adult Metabolic Diseases Clinic is recommended. Your family member will receive the same assessment by our clinic staff you received and may be requested to complete similar testing. Sometimes understanding your test results helps us to determine what may be affecting other members of your family.

Testing of family members within BC will follow the same procedure as testing for you (see the section on *How is Genetic Testing Funded*). If your family member does not reside in BC, they may be advised to consult a Metabolic Clinic in their home province. In some cases, they may be required to cover the cost of collecting and/or shipping the sample to the testing lab. This testing will be discussed with you and your family member.

How is Genetic Testing Funded?

Genetic testing is funded by the province under the Medical Services Plan of BC (MSP). Some genetic tests require an application to MSP for funding; your health care provider applies for this coverage. Funding must be approved before genetic testing can begin.

What if MSP does not approve funding?

When MSP does not approve funding, you can choose to pay for the test yourself. The cost of one genetic test can range from under a \$100 to over \$2000. The cost depends on what type of genetic test is needed. It will cost more if more than one test is needed or if multiple family members must be tested.

What Will Happen to My Sample After the Test?

Genetic Research

What happens to your sample after the genetic test will depend on the laboratory. Some laboratories may discard your remaining sample after a period of time (usually 2-5 years) but other laboratories may maintain your sample for future testing for quality assurance (to ensure the test results produced are accurate) or use your left over sample for research or education purposes.

Research testing on a sample is used to:

- find unknown genes
- learn how the genes work
- study about the genetic conditions

Information on research which uses your sample may not be sent back to you or your health care provider. Your name and other personal information will not be revealed in any research project unless you have provided specific consent to allow the publication of your name.

Providing Consent

Genetic testing is a voluntary procedure, which means **only you** can decide to get tested or not. Your physician or clinic nurse will discuss with you the procedure, benefits, and limitations of genetic testing before you make your final decision.

You will be asked to sign a **consent form** if your sample, with personal identifying information, is sent to laboratories out of the country, such as the United States. No health record will be sent with the sample. You may also be asked for permission to contact other family members who are at risk for the genetic disorder.

Who Can Help Me Learn More About Genetic Testing?

Please contact your health care provider for any further questions or concerns about genetic testing. They are the best resource to help you understand genetic testing.

For more resources, you can visit:

Genetics Home Reference

<http://ghr.nlm.nih.gov/handbook>

National Human Genome Research Institute

Genetic Information Non-discrimination Act (GINA)

<http://www.genome.gov/10002328>

National Human Genome Research Institute

Genetic Testing

<http://www.genome.gov/Pages/Health/PatientsPublicInfo/GeneticTestingFactSheet.pdf>

Genetics and Rare Diseases Information Centre

<http://rarediseases.info.nih.gov/GARD>

Medline Plus

<http://www.nlm.nih.gov/medlineplus/geneticTesting.html>

National Cancer Institute

<http://www.cancer.gov/cancertopics/UnderstandingCancer/genetesting>

Definitions

- **Autosomal** means the mutated gene that is responsible for the genetic disorder is located on one of the 22 pairs of *autosomes*.
- **Autosomes** are non-sex determining chromosomes. There are 22 pairs of autosomes.
- **Autosomal dominant conditions** are genetic conditions that are fully expressed in people who have just one copy of the mutant gene.
- **Autosomal recessive conditions** are genetic conditions that are fully expressed when the person has two copies (one copy from each parent) of the mutated gene.
- **Carriers** are people who have changes in their genes but do not have the disease. They have a chance in passing their changed gene to their children.
- **Chromosomes** are long strands of DNA tightly packaged together in the cells of the body. Human cells normally have 23 pairs of chromosomes. Half of each pair is inherited from the mother and the other half from the father.
- **Deletion** is a *structural abnormality* of a chromosome when a section of the chromosome is missing or deleted.
- **DNA or Deoxyribonucleic Acid** is the hereditary material in humans and all other organisms (Genetics Home Reference, 2011d). Like a blueprint, it has all of the instructions and information to build a person.
- **Dominant** is genetic condition that only requires one mutated gene to be expressed.
- **Duplication** is a *structural abnormality* of a chromosome when an extra copy in a section of the chromosome is made.
- **Enzymes** are proteins that speed up a biochemical reaction.
- **Genes** are found in all of the cells, tissues, and organs of our bodies and are made up of *DNA* which contains many instructions for specific activities that help our bodies grow and function. Genes also have the information that make you look the way you are.
- **Gene inheritance** is when genes are passed from parent to child.
- **Gene mutation** is a change in the DNA spelling that causes a change in the gene. A gene mutation may cause a genetic disorder that could be passed from parent to child.
- **Genetic disorder** is a disease caused by a change in our DNA spelling. This change causes our genes to no longer work.
- **Genome** is a person's complete set of DNA and it displays all of the genes of that person. Genomes carry all of the information needed to build and maintain a living person (Genetics Home Reference, 2011d).
- **Heteroplasmic cells** are cells of the human body that contain mutations in the mitochondrial DNA.
- **Homoplasmic cells** are normal cells of the human body, which contain normal mitochondrial DNA.
- **Inversion** is a type of structural abnormality of a chromosome which happens when a section of the chromosome breaks off and turns upside down before it is reattached. The genetic material is then inverted.
- **Maternal inheritance** is a type of inheritance which involves passing down genes in the mitochondrial DNA from the mother to the child.
- **Mitochondria** are tiny, cucumber-shaped "factories" that turn the food we eat into energy that are used in all body cells.
- **Monosomy** occurs when a person is missing a chromosome from a specific pair.

- **Nucleus** is the control centre of a cell and it is where genetic material such as your DNA and chromosomes are found.
- **Numerical abnormalities** are errors in the number of chromosomes
- **Proteins** are building blocks of cells that make tissues of the body.
- **Recessive** is a genetic condition that requires two copies (one copy from both parents) of the mutated gene to be expressed.
- **Reciprocal translocation** is a type of structural abnormality in a chromosome that occurs when sections from 2 different chromosomes have been exchanged.
- **Robertsonian translocation** is a type of structural abnormality in a chromosome that occurs when an entire chromosome attaches itself to another chromosome.
- **Ring** is a type of structural abnormality in a chromosome that happens when a section of the chromosome breaks off and forms into a ring. It can happen with or without losing any genetic material.
- **Structural abnormalities** are errors in the sequence of chromosomes that can be caused by deletion, duplication, translocation, and/or inversion.
- **Translocation** is a type of structural abnormality of a chromosome when a section of one chromosome is transferred to another chromosome. There are 2 types of translocations, *reciprocal translocation* and *Robertsonian translocation*.
- **Trisomy** means there is more than two chromosomes of a specific pair.
- **X-chromosome** is the sex determining chromosome.

- **X-linked** means the mutated gene that is responsible for the genetic condition is located on the X-chromosome (one of the sex determining chromosomes).
- **X-linked recessive conditions** are genetic conditions that are caused by mutations in the genes on the X-chromosome.
- **X-linked dominant conditions** are genetic conditions that will only require one copy of the mutant gene to express signs and symptoms of the genetic disease. X-linked dominant disorders are caused by mutations in genes on the X chromosome.

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