Informed Consent for Genetic Testing

What Michigan Patients Need to Know Before Getting a Genetic Test

MDHHS
Michigan Department of Health & Human Services

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Genetic testing has become important in many areas of health care. Since 2000, the state of Michigan has had a law that requires health care professionals to fully inform people who are offered certain genetic tests about the benefits, risks, and limits before testing is done. This booklet was created by the Michigan Department of Health and Human Services to help answer questions about genetic testing and to help meet the requirements of the law. The goal of this booklet is to make sure you have enough information about genetic testing to decide whether you want to have a test.

The booklet has a glossary* (list of definitions) to help you understand genetics-related terms. The booklet also includes a model consent form that contains the minimum information that a person having a genetic test must be given before testing, as required by Michigan law. Your health care provider is responsible for giving you this information and having you give signed consent for genetic testing before you have a test. Your provider may use the form included in this booklet ("Consent to Obtain a Specimen for Genetic Testing") or a similar one.

Reading this booklet may help you think of questions to ask your health care provider about genetic tests you are offered.

Genetic testing is your choice (voluntary). Only you can decide if you want to be tested. You may want more information before deciding what to do. People often have questions about the condition(s) for which testing is being considered. They may also have questions about other issues such as whether insurance covers testing. Your health care provider will explain the purpose of the test and try to answer your questions.

If after reading this booklet you still have questions, be sure to ask your health care provider (doctor, nurse, nurse practitioner, physician assistant, genetic counselor) before you agree to have the genetic test being offered to you.

***Glossary terms are highlighted in a bold, blue font throughout the booklet.**

1. What is a genetic test?

A **genetic** test is any laboratory test used to look at a person's genetic makeup. Everyone is born with a unique genetic makeup. Sometimes variants within a person's genes may cause or contribute to a medical condition, like a **birth defect** or an increased chance of developing cancer.

Depending on the specific genetic test ordered, the test may detect one genetic condition or many genetic conditions. Your health care provider should talk to you about the condition(s) for which you are being offered testing as part of the **genetic counseling** that occurs before genetic testing.

Genetic tests are usually performed using blood, saliva (spit), cheek **cell** or skin cell samples. When testing a pregnancy, **chorionic villus**, **amniotic fluid,** or a sample of the mother's blood (which includes a small amount of the baby's genetic information) can be used. Genetic tests can also be performed on embryos (fertilized eggs). These tests may look at a person's **DNA**, **RNA, proteins,** or other substances in **cells** that can indicate a genetic condition.

2. What is the purpose of genetic testing?

There are different reasons for doing **genetic** tests. A genetic test can be done to:

- Confirm a diagnosis that is already suspected based on personal and family medical history and/or a medical exam. (**Diagnostic Test**)
- Determine the chance a healthy person is at risk to develop a disease or condition in the future. (**Predictive Test**)
- Determine the chance a healthy person will develop a disease or condition in the future. (**Presymptomatic Test**)

- Determine **carrier status**. A carrier has a change in a specific **gene** or genes that increases their chance of having a child with a genetic disease or **birth defect**. (Carrier Test)

3. How effective is genetic testing?

The effectiveness of a **genetic** test depends on many factors including the purpose of the test (why it is being ordered) but also its potential benefits, risks, and limitations, as described below.

4. What are the limitations of genetic testing?

There is no one test that detects all **genetic** conditions. Genetic testing is very accurate, but it has some limits, including the following:

Performing the right test

Your health care provider must have accurate information about your personal medical history and family history to figure out which genetic test(s) to order and to correctly interpret what the results mean. If you are being tested for a condition that runs in your family, but the test used is not the correct test, your result will not accurately determine your risk for the condition.

Also, tests that depend on having samples from family members may be interpreted incorrectly if there is non-paternity (named father is not the biological father of a child) or if the true bloodlines in a family are not known.

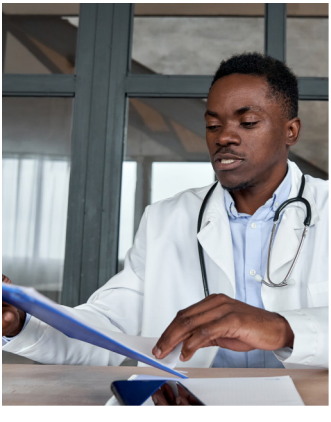
Laboratory processing

All certified (clinical) laboratories have strict rules for handling samples from the time the sample is received to the reporting of the results. In rare cases, problems may occur in handling the sample, which might lead to incorrect results. Research labs and **direct-to-consumer (DTC)** testing labs may not have the same strict rules that clinical labs have, which may increase the chance for sample mix-ups and other errors.

Interpreting what a test result means

Below is a list of possible limits to understanding what a genetic test result could mean for your health and that of your family:

- A genetic test may identify a **gene change** for which the impact on a person's health is currently unknown (it may or may not cause the condition). This is called a **variant of uncertain significance (VUS)**. It may take months or years to figure out what having a VUS means for one's health.
- A genetic test may find a gene change that confirms a suspected diagnosis (**diagnostic testing**), but the result may not give information about how severe the condition will be.
- A genetic test may identify a gene change that indicates an increased chance for developing a condition in the future (**predictive testing**), but it cannot predict whether a person will actually develop the condition, or when symptoms will appear.
- A genetic test may find a gene change that shows a healthy person will eventually develop a condition (**presymptomatic testing**), but it cannot tell exactly when the condition will develop.
- A genetic test may show a normal result (no gene change), but this may not rule out a person's chance of having a genetic condition. The test may not find all the gene changes that can cause the condition or there may be other genes that can cause the condition that were not tested or are still unknown.



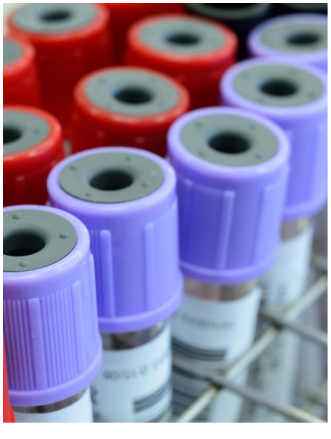
It is important that you discuss the limits of genetic testing with your health care provider so that you are fully informed about what the results might mean.

5. What are the benefits of genetic testing?

There are several possible benefits of **genetic** testing. For example:

- Knowing about an increased chance of developing a condition might lead a person to choose medical treatments that reduce their risk or help identify the condition at an early, more treatable stage.
- Having knowledge can empower a person and family members to make important life planning decisions, even if a cure is not available at that time.
- Having a specific diagnosis could qualify a person to enroll in research studies, which may lead to new treatments.
- Knowing about a certain **gene** might provide important health information for other members of a person's family.
- Confirming a genetic diagnosis can give insurance companies the information they need to decide whether to cover **screenings** or treatments for the condition.
- When parents find out that they both have a change in the same gene (**carrier** testing), this information can help with family planning.

6. What are the risks of genetic testing?



There are also several possible risks of genetic testing. These include:

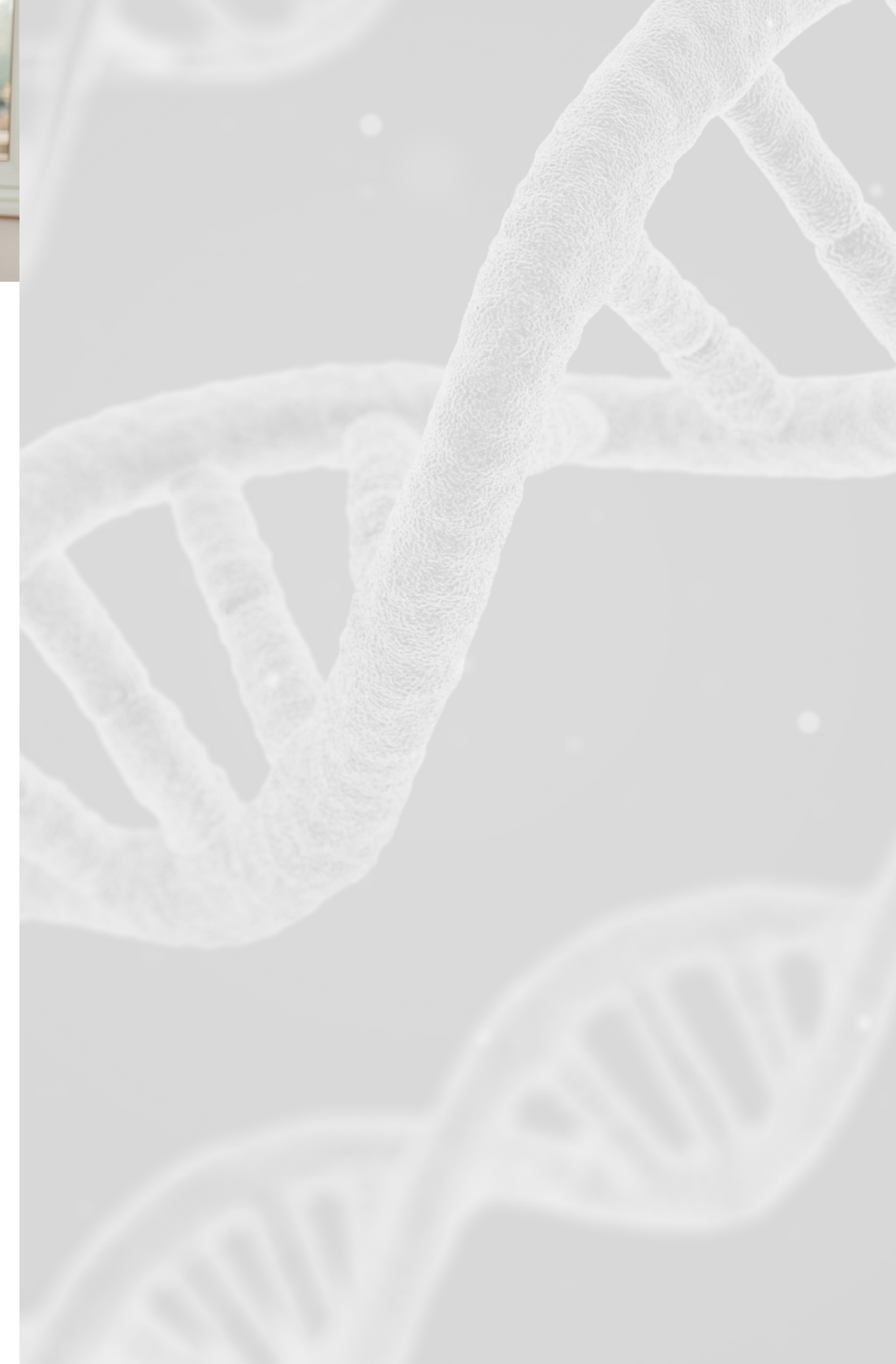
- **Physical risks.** The physical risk of testing is usually small, often not more than having a blood sample drawn. If your test involves any other type of sample, the health care provider

collecting the sample, or a professional they work with, should explain the risks before you decide to have the test.

- **Personal psychological and social risks.** These risks are related to how the results might change a person's life or views on life. The decision to have genetic testing can be stressful. A person may have emotional reactions to learning that they do or do not carry a **gene change** for a certain condition. For these reasons, meeting with a therapist may be suggested before some genetic tests are performed.

- **Familial psychological and social risks.** These risks are related to how the genetic test results might affect family relationships. A person who decides to have genetic testing needs to think about whether to tell other family members. Sometimes the test result for one family member can disclose information about the genetic makeup of other relatives, even if they have not been tested. Also, a genetic test may show unexpected relationships, such as non-paternity (a different biological father).

- **Economic risks.** Genetic testing may affect a person's ability to obtain health, life, disability or long-term care insurance. It could also affect the ability to obtain or keep a job. There are both state and federal (U.S.) laws to help protect people from genetic discrimination. Many of these laws prevent the use of genetic information by health insurers and employers but there are some limits. Life, disability, and long-term care insurance are usually not covered by these laws.



CONSENT TO OBTAIN A SPECIMEN FOR GENETIC TESTING

PATIENT LAST NAME: FIRST NAME:

(Please Print)

DATE OF BIRTH:

HOSPITAL/ ID NUMBER:

ORDERED BY:

LABORATORY NAME, CITY AND STATE:

SAMPLE TYPE

- ☐ Amniotic fluid
- ☐ Blood
- ☐ Saliva or cheek swab
- ☐ Chorionic villus sample (CVS)
- ☐ Skin
- ☐ Tissue block
- ☐ Other _____

GENETIC TESTING REQUESTED FOR:

_____

(name of condition)

The intended purpose is (check all that apply):

- ☐ Carrier status
- ☐ Diagnostic
- ☐ Predictive
- ☐ Prenatal
- ☐ Presymptomatic
- ☐ Screening
- ☐ Other _____

1. I have been informed about the nature and the purpose of this genetic test.
2. I have received an explanation of the effectiveness and limitations of this genetic test.
3. I have discussed the benefits and risks of this genetic test with my physician and/or other health care professional. I understand some genetic tests can involve possible medical, psychological or insurance issues for my family and me.
4. I understand the meaning of possible test results and have been informed how I will receive the result.

5. I have been informed that genetic testing can sometimes reveal incidental or secondary findings- results that are not related to the purpose of testing. I have discussed with my health care professional if and/or how such results will be shared with me. I understand that it is up to me to decide whether I want secondary results reported back to me and what secondary results I want reported.
6. I have been informed who may have access to my biological sample, and that any leftover sample may be retained by the laboratory.
7. I have been informed who may have access to my genetic test result, which is part of my confidential medical record.
8. My questions have been answered to my satisfaction.
9. I understand that this consent form is intended to be used together with the patient information booklet that contains important information explaining the above eight items. I have read both this consent form and the booklet. I received a copy of the form and booklet for my records.

I consent to have a sample taken for genetic testing on the above-named patient for the condition(s) listed above.

_____

Signature of Patient or Authorized Designee Date

Circle one: **Self** **Parent(s)** **Legal Guardian** **Durable Power of Attorney for Health Care**

Print Name of Physician or Authorized Delegee explaining the above information:

Signature of Authorized Person: Date:

This consent form was developed by the Michigan Department of Health and Human Services in compliance with PA 29 of 2000 and must be distributed with "Informed Consent for Genetic Testing" patient booklet. Neither may be altered nor deleted to change the meaning of specific statements above or the intent of the informed consent process.

Revised 9/21

7. What do the results of genetic testing mean?

Genetic tests can be classified as negative or positive.

Negative (no changes found in the gene(s) tested or "normal")

A negative result means a person does not have a change in the **gene(s)** tested. Usually a negative test result is good news, but there are some limits. For instance, some **genetic** conditions are caused by more than one gene and a person may not have been tested for all of these genes. Also, genetic conditions may be caused by different types of changes within the same gene, but the laboratory may not test for all of these types of changes.

For these reasons, it can be hard to interpret the meaning of a negative genetic test result. This is especially true when genetic testing is done in a healthy person before an affected family member has been tested. In such cases, a negative result in the healthy family member is called an uninformative negative result. The result does not rule out the possibility that the person tested is still at risk. That is why it is important, when possible, to first test a family member who is suspected to have the genetic condition before testing unaffected relatives. If you are considering testing, and the ideal person in the family is not available to test, talk to your health provider about the limits of interpreting your results.

The technology to identify new genes associated with a genetic condition or to detect certain types of genetic changes improves every year. If you have testing and the result is negative, you may want to ask whether a better test may become available and then check back with your health care provider periodically in the future.

Positive (pathogenic gene change detected)

- A positive test result on a **diagnostic**, **presymptomatic**, or **predictive test** means that a person has a gene change that is known to cause a specific condition, or can put a person at increased risk of developing a genetic condition. These types of gene changes are called **pathogenic or likely-pathogenic variants (P/LP variants)**. They may also be called mutations.

- A positive test result also means there is an increased chance that the person's relatives, including current or future children, could have the same genetic condition. How small or big that chance is depends on how the condition is inherited in the family. If you have testing, it is important to talk to your health care provider about what your test results could mean for your relatives and to think about how you would notify your relatives about their risks.

- A positive result on a **carrier** test means that a person has an increased chance of having a child with a specific genetic condition. In most cases, being a carrier does not increase a person's chance of having any health concerns but there are some exceptions.

If you have a positive carrier test result, it is likely that you inherited the gene change from your mother or father. As such, your relatives will also have an increased chance of being a carrier for this condition.

Variant of uncertain significance (VUS)

Sometimes it is unclear if a gene change found through genetic testing is harmful (causes or increases the chance of developing a specific condition) or **benign** (harmless). In such cases, the genetic change is called a **"variant of uncertain significance" (VUS)**. Laboratories may learn more about a VUS in the future and this may allow the lab to at some point classify the VUS as a benign or likely benign variant (harmless) or as a pathogenic or likely

pathogenic variant (associated with disease or risk of disease). For this reason, if you have testing and it shows a VUS, it is important to contact the health care provider who ordered your genetic testing once or twice a year to see if more information is available.

Incidental or secondary findings

Sometimes genetic testing reveals a finding that is unrelated to the reason for doing the test. These are called **incidental or secondary findings (ISFs)**. Examples include:

- **A risk for a genetic condition that is unrelated to the original reason for testing.** Some tests look for genetic changes across a person's genetic makeup, for example, whole exome or whole genome tests, instead of just looking at genes known to be related to the person's symptoms and family history. When these types of tests are done, occasionally a pathogenic/likely pathogenic variant (P/LP variant) will be found in an unrelated gene. In addition, the American College of Medical Genetics and Genomics recommends that when such tests are done, laboratories look for P/LP variants in a specific set of actionable genes. Actionable genes cause genetic conditions, but early identification and treatment can improve a person's health outcomes.
- **Non-paternity.** Finding out the person designated as the father is not the biological father of the person having testing.
- **Consanguinity.** Finding out the biological parents are closely-related by blood.

Genetic testing laboratories have different ways of reporting ISFs to patients and providers. How they are reported depends on a number of factors including laboratory policies, whether knowing the result will change a person's medical care, and a person's preferences. It is important for your health care provider to discuss how you want any potential ISFs handled during your pre-test **genetic counseling**.

Overall, before you decide to have genetic testing, make sure you understand what a negative, positive, or VUS result will mean by discussing your situation with your physician, nurse, nurse practitioner, physician assistant or genetic counselor. Also, speak to your health care professional about the possibility of ISFs results and how they will be handled.

8. What will happen to my sample after the genetic test is completed?

You have the right to know what will happen to any remaining sample. As part of **informed consent**, your health care provider should discuss this with you before **genetic** testing. The consent form may allow you to choose whether you are willing to have your sample used for other purposes, such as **research**.

9. Who has access to my sample and information from the test?

Genetic tests are handled in a confidential manner, like other personal health information. The person collecting your sample, and people in the laboratory performing the test, will handle your sample. The health care provider who ordered the test will receive the results and should let you know how you will be informed about these results. Your medical record and test results are confidential. Your written permission is required before your medical record is released to anyone else.

Genetic information is familial information. Sometimes your test results can provide your family members with information about their genetic risks that could be life-saving. Therefore, it is important for you to thoughtfully consider sharing your results with your family, especially if the condition you are being tested for is preventable. You can discuss with your health care provider ways that you can share your information with your family.

10. What is the cost of genetic testing?

The cost depends on the test performed. Usually the cost can range from a few hundred to several thousand dollars. Before having testing, ask about the cost of the test, whether it is covered by insurance, and if you will be responsible for any or all of the bill. Whether a test is covered depends on the test, the reason the test is being ordered, and your insurance plan.

Some **genetic** tests are regularly covered by most medical insurance plans. However, some are not. Also consider whether you have a deductible for your health insurance and whether you have met the deductible for the year.

11. Where can I get more information about genetic testing and related services in Michigan?

You can ask your physician, **genetic** counselor, or other health care provider. You can also call the Michigan Department of Health and Human Services toll-free at 866-852-1247 or visit the MDHHS Public Health Genomics Program website at **Michigan.gov/Genomics**.

Glossary of Genetic Terms

Amniotic fluid: The fluid around an unborn fetus; it contains fetal cells that can be used for genetic testing.

Benign or likely benign variant: A gene change (variant) that is not thought to cause or increase a person's risk of developing a disease or condition.

Birth defect: A physical difference in a baby that is present at birth. The physical difference may occur alone or be associated with developmental disabilities.

Carrier status: The knowledge of whether a healthy person has a disease-causing gene change (mutation) that could be passed down to their children. Carrier status is often determined by genetic screening.

Cell: The basic unit of all living things, including humans.

Chorionic villus: Cells found in the placenta during pregnancy. These cells contain the same genetic information as the developing fetus and can be used for genetic testing.

Diagnostic testing: A test to confirm the presence or absence of a specific disease or condition.

Direct-to-consumer (DTC) testing: Testing that a person orders on their own without a health care provider. Results are returned directly to the person.

DNA (Deoxyribonucleic Acid): A particle or material inside a cell that carries the genetic information needed to operate that cell, make tissues and control organ systems.

Fetus: An unborn baby from about eight weeks after conception until birth.

Gene: A subunit of DNA that contains the message for a cell product, typically some type of protein. Humans have approximately 20,500 genes.

Gene change (mutation or variant): A change in the genetic structure or DNA. Some gene changes increase a person's risk of developing a disease or condition (pathogenic) while others do not increase risk for health conditions (benign).

Genetic: A trait or condition determined by one or more genes.

Genetic counseling: The process of helping people understand a genetic condition and how to adapt to the implications of having the condition. The process involves: assessing a person's chance of having a genetic condition based on family history, medical history and genetic test results; providing education about the condition(s) and what it could mean to the family; helping the patient and family make informed choices about the use of genetic information based on the family's values and beliefs; and helping the family adapt to living with the

condition or risk for the condition.

Germline genetic test: A test that looks for genetic variants that a person inherited from their parents and can pass down to their children.

Incidental or secondary findings (ISF's): A genetic test result that provides information unrelated to the reason for the test. This includes finding a change in a gene that is not related to a person's symptoms, learning about non-paternity (different biological father), or learning about consanguinity (parents are closely related by blood).

Informed consent: A person's agreement to allow a medical test, treatment or procedure based on a full understanding of all the facts necessary to weigh the benefits, risks, and limitations.

Pathogenic or likely pathogenic variant (P/LP variant): A gene change (variant) that likely caused or increases the risk of a person developing a disease or condition.

Predictive testing: A genetic test to determine if a person has one or more gene changes that increase the risk of developing a certain disease or condition at some time in the future.

Presymptomatic testing: A genetic test performed before the onset of any symptoms to determine if a person has a gene change that will eventually cause a certain disease or condition.

Protein: A particle in living things that is needed to support body structure, function and regulation. Proteins are made according to the DNA sequences in genes.

Research genetic testing: Testing that is usually done to find out whether certain genes cause a condition, how the genes work, and/or what risks may be associated with having a variant in the gene(s). Research genetic test results may not be shared. If shared, the results should always be confirmed in a clinical lab.

RNA (Ribonucleic Acid): A particle in the body that is similar to DNA. It plays an important role in making proteins and in other cellular activities.

Screening: The process of looking for a particular gene change or disease in individuals who don't exhibit any signs or symptoms.

Somatic genetic test: A test that looks for genetic changes in cancer cells that caused the cancer to develop. These genetic changes are acquired, not inherited, and are only present in the cancer cells.

Variant of uncertain significance (VUS): A gene change (variant) for which there is not enough information to know whether it will cause or increase a person's risk of developing a disease or condition.

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To order more copies of the booklet please e-mail genetics@michigan.gov

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