

Informed Consent for Genetic Testing

Patient Education Information for Use with the Michigan
Model Consent Form for Genetic Testing

Many new advances are occurring in medicine, especially in the field of medical genetics. This booklet was prepared by the Michigan Department of Health and Human Services in compliance with Michigan law to help answer your questions about genetic testing. Our goal is to make sure you are well informed before you decide to give a sample for testing.

The booklet includes a glossary to help you understand genetics-related terms. The booklet also includes a model consent form that contains the minimum information an individual undergoing a genetic test must understand as required by Michigan law. It was developed to help you and your health care provider discuss information that you should understand before consenting to a genetic test. Your health care provider is responsible for ensuring that the consent form that you sign complies with Michigan law.

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

If you still have unanswered questions, be sure to ask your physician, nurse, nurse practitioner, physician assistant, or genetic counselor before you agree to have the genetic test being offered to you.

1. What is a genetic test?

There are many different types of genetic tests. A genetic test is any analysis used to look at a person's genetic makeup.

Everyone is born with a unique set of genes. Sometimes a person's genes may cause a disease or birth defect. Genes can also cause certain physical features, or an increased chance of developing a certain condition.



Depending on the type of genetic test ordered, the test may detect a single genetic condition or multiple genetic conditions. Your health care provider should inform you about the condition(s) for which you are being offered testing as part of the genetic counseling that occurs before genetic testing.

A genetic test is considered a **predictive** test if a result indicates a person has an *increased chance* of developing a disease or condition in the future, but developing the disease is not a certainty. A genetic test is considered a **presymptomatic** test if a result indicates a person *will eventually develop* a disease or condition. A genetic test is considered a **diagnostic** test if a result confirms a suspected diagnosis.

Genetic tests are usually performed using a blood sample, specifically white blood cells. Tissue or cheek cell samples also may be used. 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. The test may examine a person's **DNA, RNA, proteins**, or other chemicals 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.

- Determine the chance a healthy person is at risk or will develop a disease or condition in the future.
- Determine **carrier status**. A carrier has a change in a specific gene or genes that increases his or her chance of having a child with a genetic disease or birth defect.

3. What are the limitations of genetic testing?

There is no one test that detects all genetic diseases. Genetic testing is highly accurate, but there are some limitations, including the following:

■ Performing the correct test:

Your health care provider must have accurate information about your personal medical history and family history to determine which genetic test(s) to perform and correctly interpret the results. If you are being tested for a disease that runs in your family, but the test used is not for the condition your relative(s) actually had, your own result will not give accurate information about whether you have or are at risk for the condition. Tests that depend on having samples from relatives 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 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.

■ Implications of results:

The significance of a genetic change for your health/your family's health may not be known at the time of testing. A genetic test result may reveal a change in a gene associated with having or being at risk for a genetic condition. However, there can be limits to the information provided when a gene change is found, such as:

- A gene change that confirms a suspected diagnosis (diagnostic testing) may not give information about how severe the condition will be.

- A gene change that indicates an increased risk for developing a condition in the future (predictive testing) cannot predict with certainty whether a person will actually develop the condition, or when symptoms will appear.
- A gene change that shows a healthy person will eventually develop a condition (presymptomatic testing) does not tell exactly when the condition will occur.

It is important that you discuss your genetic test results with your health care provider so that you are fully informed about what the results mean for you and your family.

4. What are the benefits and risks of genetic testing?

■ Benefits:

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

- Finding an increased risk of disease might lead a person to choose preventive or therapeutic medical treatments that reduce the chance of developing the disease.
- 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 a person's family.

■ Risks:

There are also several possible risks of genetic testing. For example,

- The physical risk of testing is usually minimal, typically not more than providing a blood sample. If your test involves any other type of sample, the physician performing the procedure, or a designated representative, should explain the risks before you decide to have the test.



- Potential psychological and social risks of testing are related to how the results might change a person's life. The decision to have genetic testing can be stressful. You may have emotional reactions to learning that you 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.
- Additional psychological and social risks include the impact of testing on family relationships. A person who decides to have genetic testing needs to consider whether to tell other family members. Sometimes the result for one family member can disclose information about the genetic makeup of other relatives, even if they have not been tested. Furthermore, a genetic test may reveal unexpected relationships, such as non-paternity (a different biological father).
- There are possible economic risks of genetic testing that 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.
- Genetic test results are part of your medical record. If a genetic test is performed, your insurance company may have access to the result.
- There are both state and federal laws to help protect a person. The laws prevent the use of genetic information by health insurers and employers. Please visit <https://migrc.org/PublicHealth/PolicyandLaw.html> for direct links to help you find further information about such laws.

5. What do the results of genetic testing mean?

Genetic tests are often a valuable tool, but sometimes do not provide a definite answer. Most often, results can be classified as negative or positive.

■ Negative or “normal”:

A negative or normal result means you do not have a change in the gene(s) tested. Usually a negative test result is good news, but there are some limitations. Some clinical conditions can be caused by more than one gene and the laboratory may not have

tested for all of these genes. Other conditions may be caused by different types of changes within the same gene, but the laboratory may not test for all of the types of changes. This means a negative test does not always rule out the presence of every gene change that could cause a disease. You still might have a change in a gene, even if your test is normal.

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

■ **Positive or “abnormal”:**

A positive or “abnormal” test result means you carry a gene change that is known to cause a specific condition, or can put you at increased risk of developing a genetic condition. Not all gene changes lead to symptoms.

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

A positive test result also indicates that you have an increased chance of having a child with the same genetic condition. How small or big that chance is depends on how the condition is inherited in the family. If you have children or are considering having children, it is important to talk to your health care provider about what your test results mean for them.

■ **Inconclusive:**

Sometimes a test result falls in between the normal and abnormal range. In such cases, a genetic change, called a **variant** (sometimes referred to as ‘variants of uncertain significance’), may be identified but the laboratory does not have enough information to determine whether or not the change is associated with a genetic condition/risk. Laboratories may learn more about a variant in the future and this may allow the lab to eventually classify the variant as a normal (negative) or abnormal (positive) test result. For this reason, if your testing reveals a variant, it is important to contact your health care provider periodically to see if any more information is available.

CONSENT TO OBTAIN A SPECIMEN FOR GENETIC TESTING

PATIENT LAST NAME: (Please Print)	FIRST NAME: MI:
DATE OF BIRTH: / /	HOSPITAL/ ID NUMBER: GENETIC TESTING REQUESTED FOR: _____
ORDERED BY: LABORATORY NAME, CITY AND STATE: _____	The intended purpose is (check all that apply): SAMPLE TYPE <input type="checkbox"/> Amniotic fluid <input type="checkbox"/> Blood <input type="checkbox"/> Cheek swab <input type="checkbox"/> Chorionic villus sample (CVS) <input type="checkbox"/> Skin <input type="checkbox"/> Tissue block <input type="checkbox"/> Other _____
<input type="checkbox"/> Carrier status <input type="checkbox"/> Diagnostic <input type="checkbox"/> Predictive <input type="checkbox"/> Prenatal <input type="checkbox"/> Presymptomatic <input type="checkbox"/> Screening <input type="checkbox"/> 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 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.

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■ **Secondary Findings:**

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

- 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.
- Other unexpected findings, such as identifying a genetic risk unrelated to the condition for which testing was being performed. Some tests look for genetic changes across a person's genetic makeup, rather than just looking at specific genes known to be related to the person's symptoms/ family history. When this type of testing is done, there is a possibility that it might reveal a risk for a genetic condition that is unrelated to the original reason for testing.

Genetic testing laboratories have different ways of reporting these types of secondary findings 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 your preferences. It is important for your health care provider to discuss how you want any potential secondary findings handled during your pre-test genetic counseling.

Overall, before you proceed with genetic testing, make sure you understand what a negative, positive, or inconclusive test 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 secondary results and how they will be handled.



6. 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 the informed consent, your health care provider should discuss this with you before genetic testing. The consent may allow you to say whether you are willing to have your sample used for other purposes, such as research.

7. 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. 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 important information about their genetic risks. Therefore, it is important for you to be willing to share your results with your family, especially if the condition you are being tested for is preventable. You should discuss with your health care provider ways that you can share your information with your family.



8. What is the cost of genetic testing?

The cost depends on the test performed. Typically the cost can range from a few hundred to several thousand dollars. Before having testing, ask about the cost of the test and whether it is covered by insurance. Find out if you will be responsible for any or all of the test bill. Genetic tests are not always covered by medical insurance, and you may need to pay some or all of the cost. Also consider whether you have a deductible for your health insurance and whether you have met the deductible for the year.



9. 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 1-866-852-1247 or visit www.migrc.org.

GLOSSARY OF GENETIC TERMS

Amniotic fluid:

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

Birth defect:

An abnormal condition that occurs before or at birth. Birth defects can cause physical and developmental problems that may require special medical care or therapy.

Carrier status:

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

Cell:

A small membrane-bound compartment, filled with chemicals; the subunit of all living things, including humans.

Chorionic villus:

Cells contained in the tissue of the placenta in early pregnancy. The tissue contains the same genetic information as the developing fetus.

Diagnostic testing:

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

DNA (Deoxyribonucleic Acid):

A large molecule that carries all of the genetic information needed to operate a cell, make tissues and control organ systems.

Enzyme:

A protein that speeds up a biochemical reaction.
Enzymes are critical to cell functions.

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 such as an enzyme. Humans have approximately 20,500 genes.

Gene Change:

A change in the genetic structure or DNA. This is also called a 'mutation'.

Genetic:

A trait or condition determined by one or more genes.

Genetic counseling:

The process of helping people understand the medical facts about a genetic condition and helping them adapt to the implications of the condition. The process involves: assessing a person's chance of a genetic condition based on family history, medical history information and genetic test results; providing education about the genetic condition(s) for which they are at risk 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.

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 and risks.

Molecule:

A chemical combination of two or more atoms that form a specific chemical substance; the smallest unit of a substance that displays characteristic physical and chemical properties.

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 large complex molecule essential to body structure, function and regulation. Examples include hormones, enzymes, and antibodies. Proteins are determined by the DNA sequences within genes.

RNA (Ribonucleic Acid):

A cell molecule similar to DNA. It plays an important role in making proteins and other cell activities.

Screening:

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



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