

How does genetic testing work?

Iowa Institute of Human Genetics



UNIVERSITY OF IOWA
HEALTH CARE

Iowa Institute of Human Genetics

What is a genetic test?

A genetic test looks at your DNA to find changes (variants) that cause disease or put you at greater risk to develop disease. DNA is the code our bodies use to make genes, and genes are the instructions for our bodies.

In the past, it was possible to screen, or test, only one gene at a time to try to find the cause of a disease. This method is still used for many diseases, but it can be costly and time consuming. It is now possible to screen thousands of genes at once.

Why would I have a genetic test?

A genetic test can help you and your doctor:

- Find the cause of your disease or symptoms
- See if you are at risk to develop a disease in the future
- Tell you if you could pass a disease on to your children
- Choose the best treatment for you
- Provide better counseling for you

It is always your choice if you want to have genetic testing or screening.

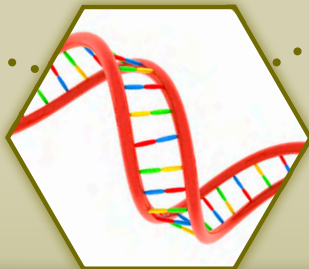
What are different types of genetic tests?

- **Clinical genetic tests:** These are tests ordered by your doctor and done in a certified lab. Results of a clinical genetic test become part of your medical record.
 - **Diagnostic test:** to find the cause of a disease or symptoms you already have
 - **Presymptomatic test:** before you have symptoms, to find if you are at risk to get a disease in the future
 - **Carrier test:** to find DNA variants that can cause disease if both parents pass these variants on to their children; carriers usually do not have symptoms of the disease
 - **Prenatal test:** a test during pregnancy to see if the baby will have a disease
 - **Newborn screen:** a test performed one to two days after a baby is born to see if the baby has certain diseases
 - **Pharmacogenomic test:** to find the best medicine and dose for you based on your DNA variants
- **Research genetic test:** You volunteer to participate in a research study, and the results are not put in your medical record.

What is the genetic testing process?



1
Your visit to
the doctor's
office



2
Prepare
your DNA



3
Sequencing
your DNA



4
Analyzing
your DNA



5
Interpreting
your DNA



6
Receiving
your results

How is a genetic test performed?

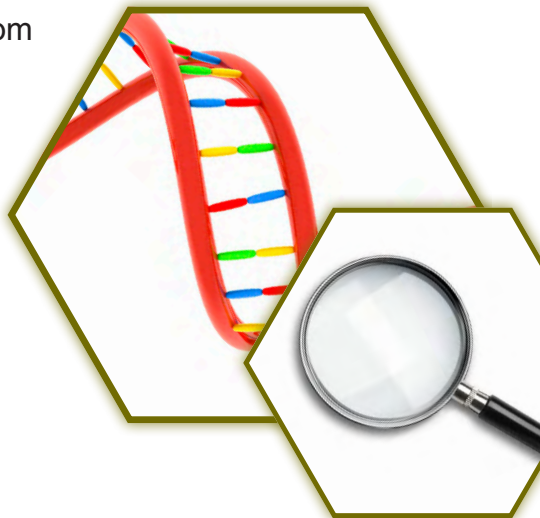
Step 1 – In the doctor's office:

- Your doctor or genetic counselor will talk to you about the test, what it may or may not tell you, insurance coverage, and risks of insurance discrimination.
- You may have to sign a consent form depending on the lab performing the test.
- Your doctor or nurse will get a sample from you. Samples are usually blood samples, but sometimes can be saliva, skin, fluid around the baby during pregnancy, or tumor.
- You may be asked to give another sample if the first sample does not work.
- The sample is sent to the lab.



Step 2 – Prepare your DNA

- The lab will get your DNA from the sample to test.
- To find the cause of a disease, the lab can use different ways to look at all or part of your DNA. The way the lab looks at your DNA will depend on the type of genetic test your doctor ordered.



Step 3 – Sequencing your DNA

- Sequencing tests read your DNA and look for variants (changes) in the DNA.
- Sequencing tests can give detailed information about:
 - Specific genes and DNA variants (targeted sequence capture)
 - All of the genes that make proteins in a person (whole exome sequencing)
 - All of the DNA in your genome (whole genome sequencing). The human genome is made of DNA and contains all of a person's genetic information (like a set of books).
- The lab may put all or part of your DNA on a machine called a DNA sequencer.
- The DNA sequencer reads your DNA and collects data known as short "reads." Each "read" represents a very tiny part of your total DNA sequence (each read is like a page in a book).



Step 4 – Analyzing your DNA

- Computers are used to put all of the pieces (reads) of your DNA together (like putting the pages of the book in the right order).
- Computers are used to compare your DNA to all of the DNA in the human body (the human reference genome).



Step 4 – Analyzing your DNA (continued)

- The lab gets a report from the computers, which lists any variants in your DNA.
 - We all have DNA changes (variants)
 - Some DNA variants cause disease (like a word spelled wrong); other variants do not cause disease (like a word with different spellings: grey and gray).
 - We do not know what every DNA variant means at this time.

Step 5 – Interpreting your DNA

- A team of experts talks about and analyzes your results. Doctors, scientists, genetic counselors, and computer experts are all on the team (like a group of people sitting down to read and talk about a book).
- To decide which variant causes the disease you are interested in, the team looks at your results (your list of variants), your symptoms, and your family history.
- The team may or may not be able to find the cause of the disease you are interested in.
- If the team finds the cause of the disease, they confirm the result with a different test.
- Some reasons why the team may not be able to find the cause of the disease can be:
 - Some parts of the DNA cannot be sequenced.
 - Not enough is known about the disease at this time.
 - The test that was performed did not look at the part of your DNA that is causing the disease, and a different test needs to be ordered.
 - The team was not given enough information about your symptoms or family history to interpret the results.



Step 6 – Receiving your results

- The lab will give the written results back to your doctor or genetic counselor. Results of clinical tests will go in your medical record. Research study results do not go in your medical record.
- Your doctor and/or genetic counselor will tell you the results and answer your questions.
- Your doctor may need to perform more medical or genetic testing based on the results of your test.
- The amount of time to get your test results can vary.



Did you know?

Every person has
3 to 4 million
DNA variants.

Helpful Websites

Iowa Institute of Human Genetics (IIHG)

Information for patients, health care providers, researchers, and students on human genetics and personalized genomic medicine

>> www.medicine.uiowa.edu/humangenetics/

National Human Genome Research Institute (NHGRI)

Information on human genetic disease and human genetic disease research, and an explanation of your rights concerning genetic discrimination

>> www.genome.gov

Genetics Home Reference

Information about how genes affect your health: what genes are, how they work, and how they can cause illnesses

>> www.ghr.nlm.nih.gov

Helpful Websites (continued)

Genes in Life

Information about genetics and how it affects you and your family, why you should talk to your health care providers about genetics, how to get involved in genetics research, and more

>> genesinlife.org

Ask for our other booklets about personalized genomic medicine, genetic counseling, exome sequencing, and pharmacogenomics.



UNIVERSITY OF IOWA
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Iowa Institute of Human Genetics

University of Iowa
285 Newton Road, 5296 CBRB
Iowa City, IA 52242
(319) 335-3688
iihg@uiowa.edu

www.medicine.uiowa.edu/humangenetics



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