



Secondary findings in genetic testing

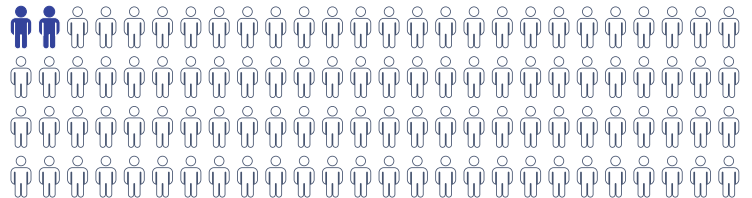
Your healthcare provider recommended exome or genome sequencing for you or your child. These tests study many pieces of genetic information (genes) at the same time to look for changes (also known as variants) that might explain your developmental and/or health concerns. With this explanation, your medical team may begin or adjust a treatment and/or management plan.

In addition to these primary findings, testing sometimes identifies genetic variants unrelated to the reason testing was ordered, called **secondary findings**.

What are secondary findings?

The American College of Medical Genetics and Genomics (ACMG) has created a list of specific “secondary finding” genes known to be associated with health conditions for which medical screening and/or treatments are available. Most of these conditions can increase the chances of developing certain cancers or heart conditions, or change the way your body breaks down, stores, and uses different substances (called metabolic conditions). Certain conditions, such as Alzheimer’s and Huntington’s disease, are not included.

Exome or genome sequencing identifies secondary findings in approximately **2% to 3%** of people. This means that testing does not identify a secondary finding in 97% to 98% of people.



Making a decision about secondary findings

Secondary findings are optional. Patients and relatives being tested can opt-in or opt-out of receiving them. If a patient decides they want to know about secondary findings as part of exome or genome sequencing, any relatives contributing a sample can also choose if they want to be checked for secondary findings, but only if they are first identified in the patient. The laboratory does not look for additional secondary findings not identified in the patient.

Deciding if you want to learn about secondary findings is personal, and there is no wrong choice.

- Some people feel this information is helpful because it might result in changes to their healthcare.
- Others feel this information may cause stress and anxiety about if, or when, they might start experiencing related symptoms.



During the consent process, you and your healthcare provider will discuss whether you want to opt-in or opt-out of secondary findings. They will help you indicate this choice on the genetic testing consent form. Any relatives being tested can also indicate their choice on the consent form.

Non-medical implications of secondary findings

In the United States, federal and state laws help prevent discrimination based on genetic test results.* The Genetics Information Non-Discrimination Act (GINA) is a federal law that prevents health insurance companies from denying or changing health insurance coverage based on genetic testing results. GINA also prevents employers from using genetic information in employment decisions but does not protect against genetic discrimination by companies providing disability, life, or long-term care insurance.

There are several groups not covered by GINA:

- Military members
- Veterans receiving care through the Veterans Administration
- Indian Health Service members
- Federal employees covered under the Federal Employees Health Benefits Program
- Individuals who work for companies with fewer than 15 employees

You can find additional information about GINA at ginahelp.org.

*Some U.S. states, specific organizations, and other countries may have policies that provide additional protection against genetic discrimination.

The test report identified a secondary finding. What are the next steps?

- ✓ The healthcare provider who ordered your genetic test will discuss the result with you. If your primary care provider is not the one who ordered your testing, it's also important to share your results with them.
- ✓ If you or your child are under the age of 18 and the secondary finding is associated with a condition that usually affects adults, there may not be any immediate changes to medical care.
- ✓ If the condition requires immediate changes to medical care, your provider might order additional non-genetic tests, such as an ultrasound or blood test. The provider who ordered the genetic testing might also refer you to a genetic counselor or specialist.

Do you have additional questions about secondary findings?

You can:

- Speak to your healthcare provider.
- Ask your healthcare provider for a referral to a local genetic counselor.
- Search for a local genetic counselor: findageneticcounselor.nsgc.org

If you still have questions, GeneDx provides genetic counseling services. Ask your healthcare provider for a referral to speak to our Patient Counseling Team.



Ready to learn more about exome and genome sequencing?

Visit GeneDx.com/patient-overview.