

Genetic Testing: The Basics



What is genetic testing?



Genetic testing looks at our DNA to find any differences that might affect our health and development.

Our DNA is like a set of instructions for our bodies. When there are differences in these instructions (also called variants), our health can be affected. There are many types of DNA differences that genetic testing looks for, like changes in how our DNA is spelled, or for extra or missing pieces .

When would someone think about having genetic testing?

If someone:

- has **signs or symptoms of a genetic condition**
- has **multiple health or developmental problems** where the cause is unknown
- has a **family history** of health conditions that might be genetic
- is **worried about their general health** and wellbeing
- wants to know **if certain medications will work** better or worse for them



How is genetic testing done?

The type of sample needed for genetic testing depends on the type of testing needed. **Usually a saliva sample, a cheek swab, or a blood sample is used.**

Saliva and cheek swab samples can be collected at a doctor's office or at home. Blood or other tissue samples (e.g. skin, tumor, etc.) will be collected in a doctor's office or lab.

Then the **samples are sent to a lab to be analyzed.** The time it takes for results depends on the type of test.



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How much does it cost? Who pays?



The cost and who pays for the test depends on the type of test, what it's for, and the patient's insurance company.

Some tests can be less than \$100, while some can cost thousands.

Insurance companies often have a set of rules to decide which tests they cover and for what reasons they will cover it. If a test is not covered, a patient may have to pay for the test themselves. Some labs offer financial assistance to help cover costs for families who cannot afford it.

How accurate is genetic testing?

How accurate the results are depends on the type of lab and testing.

To be considered **clinical testing**, a genetic test and the laboratory performing the test have to meet certain standards. These tests are proven to be accurate (correct and trustworthy) enough to be used for a patient's healthcare. Tests that can only be ordered by a healthcare provider are almost always clinically approved tests.



Non-clinical testing (e.g. direct-to-consumer 'DTC' testing and some 'at-home' testing) does not have to meet the same standards, and accuracy can vary a lot. Non-clinical tests, such as DTC tests, should not be used to make decisions about a patient's healthcare.

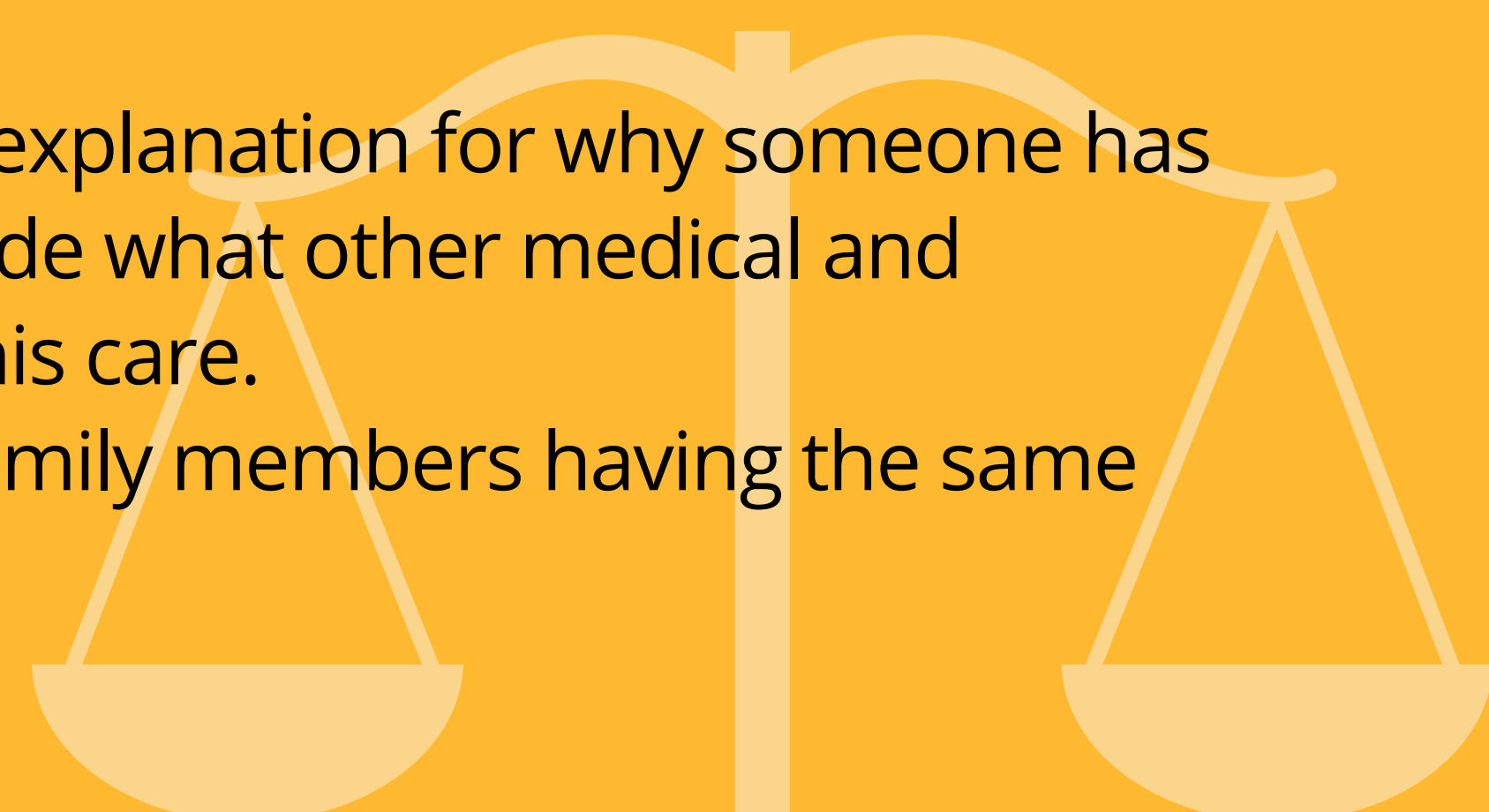
What are some benefits and risks of testing??

Benefits:

- Genetic testing can help families by finding a diagnosis or explanation for why someone has medical or developmental issues. A diagnosis can help guide what other medical and developmental care people need, and help them access this care.
- Families can find out more about the likelihood of other family members having the same types of health issues.

Risks:

- There can be a risk for whether someone is eligible for certain types of insurance, like long-term care, life, and disability. Health insurance eligibility is protected through the Genetic Information Nondiscrimination Act ([click for more information on protections and limitations of this law, like protection against employer discrimination based on genetic information](#)).
- Genetic testing may find information someone was not expecting or did not want to know.



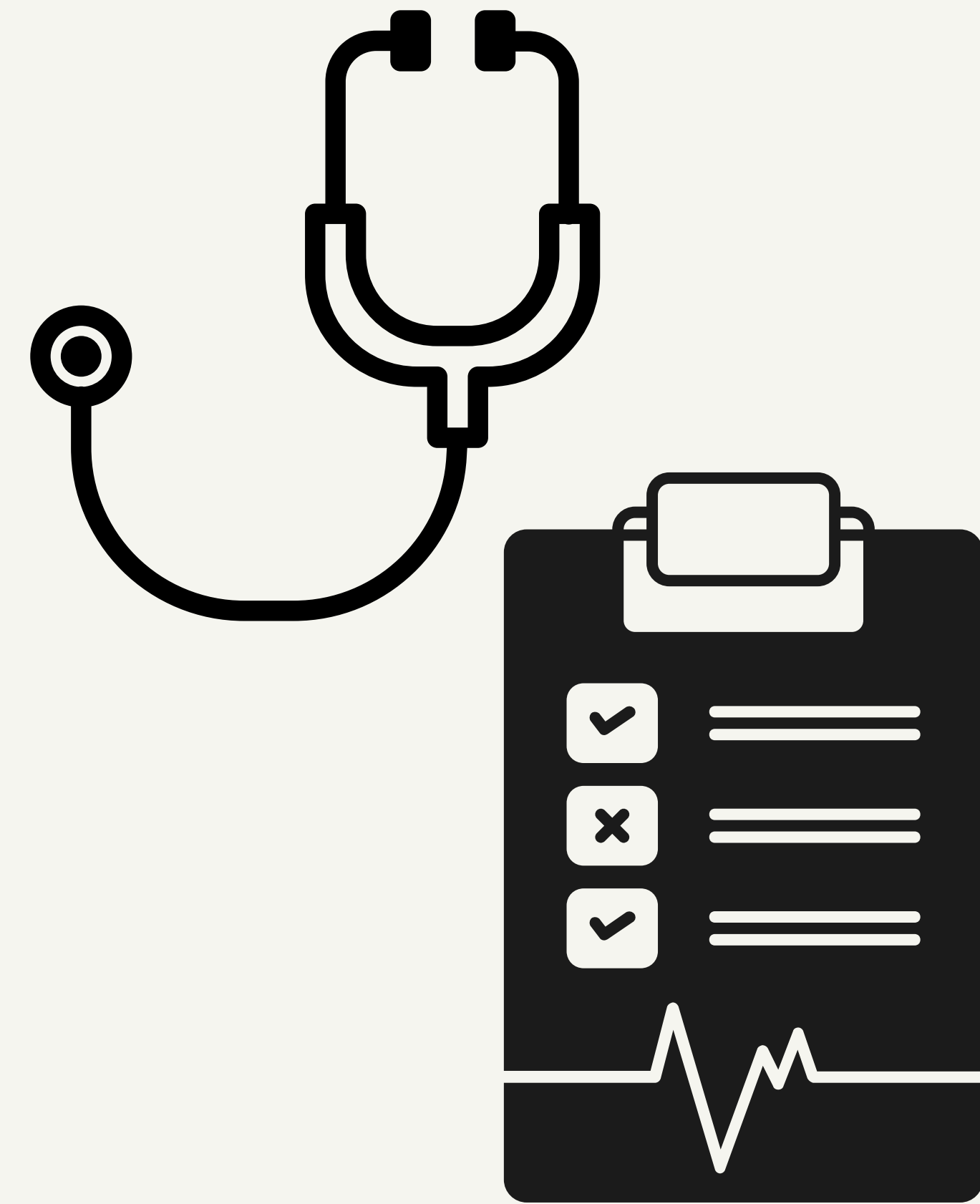
Provider Ordered vs. At-Home Genetic Testing



How can tests be ordered?

Provider ordered: A healthcare provider (e.g. doctor or genetic counselor) may order a genetic test if a patient has a personal or family history of symptoms or conditions that are known to have genetic causes.

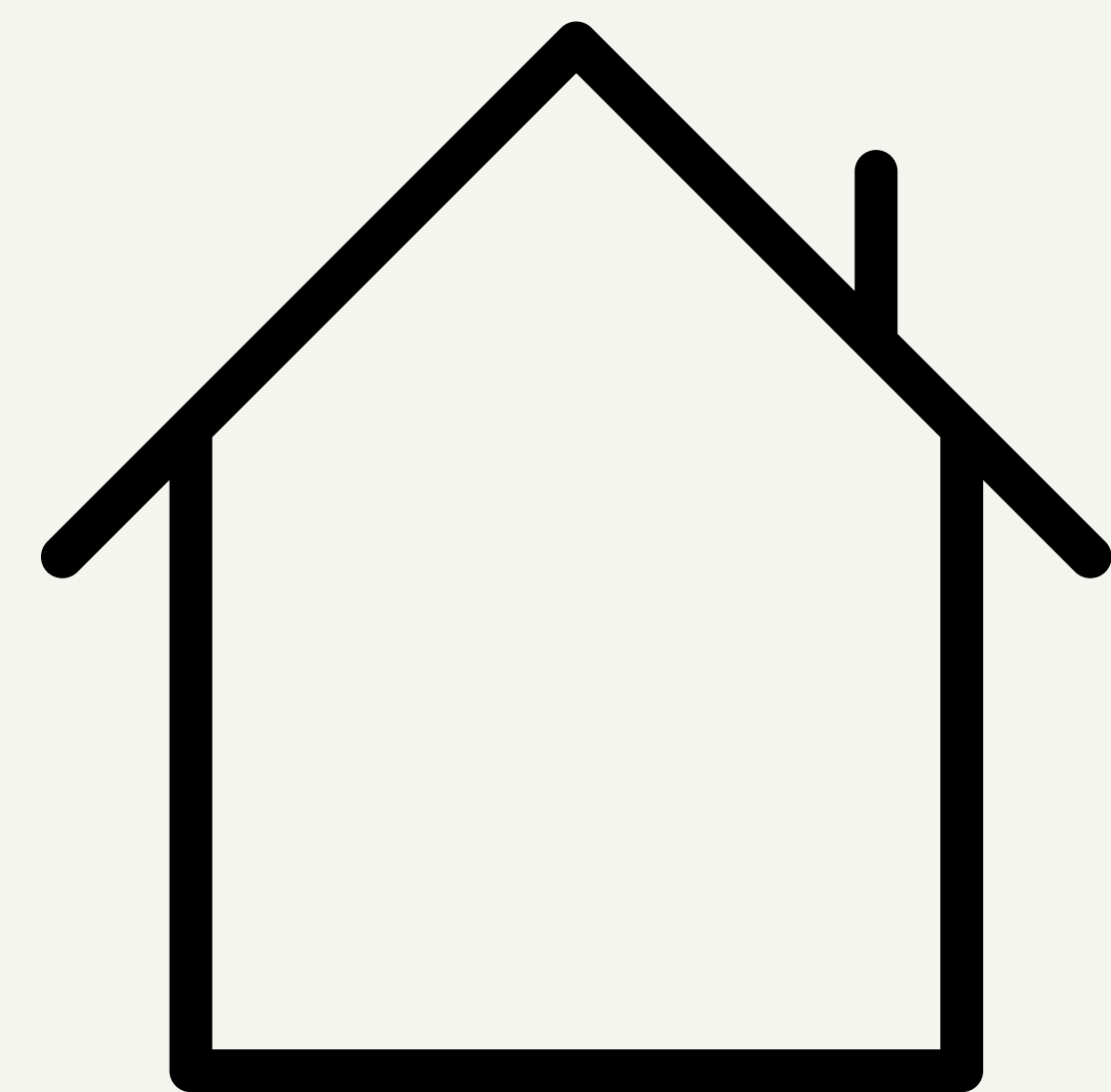
At-home: Whenever someone decides they would like to have testing, they might be able to order from labs directly. Some tests still need a provider to approve. Labs may have their own systems in place to check if the test is right for the patient.



Which types of tests can be ordered at home?

At-home testing can be clinical or non-clinical. Typically, clinical tests that can be used to guide healthcare still need to be approved by providers.

Examples of non-clinical tests are "wellness" and ancestry tests. Wellness tests might say they know how to improve your overall health based on your DNA. Ancestry tests may tell you about where in the world your ancestors came from and sometimes offer non-clinical health information.



Can I use my "raw" genetic data from an ancestry or wellness test to get clinical health information?

No. While these tests might say they give you access to ALL of your DNA and genetic information, these tests are not approved for clinical use and their results may not be accurate.

A recent study showed that almost half of "abnormal" results found using "raw data" from these types of tests were not accurate.



Provider Ordered vs. At-Home Genetic Testing



Advantages of At-Home Genetic Testing

- Usually less out-of-pocket cost
- Ability to choose test (out of the available options)
- Does not always involve a doctor's appointment or approval from another healthcare provider
- Convenience: shorter wait times, less travel required, no schedule restrictions

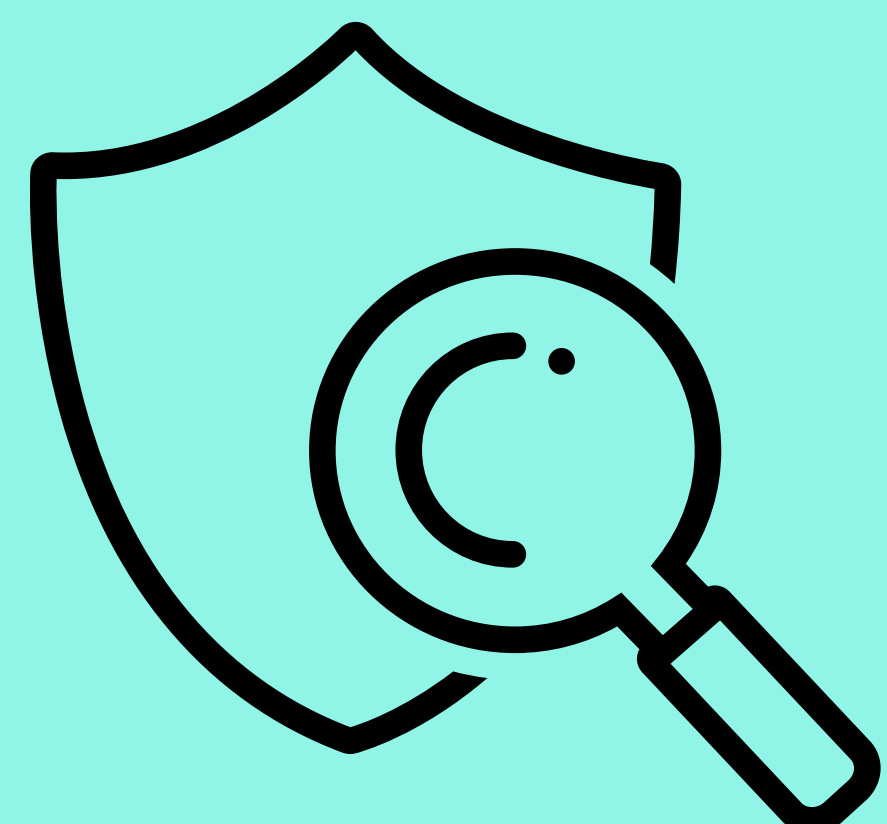


Disadvantages of At-Home Genetic Testing

- Less access to professionals that can explain the risks and benefits or answer questions about testing
- No physical exam that could help choose the right test
- Fewer test options; tests may not address patient's main worry
- Less often covered by insurance
- Often not clinically approved and results cannot be used to guide healthcare decisions
- Lack of counseling to help understand results and what they mean for the patient and their family

Is genetic testing safe/private?

It depends. All companies offering at-home genetic testing, whether clinically approved or not, should have public information about their privacy practices. Some privacy issues to think about are: selling your data to another group/company, using your data for research, and giving your information to law enforcement. Other questions to ask: "How long can they keep my data?", "How is my data protected?", or "Who has access to my data?"



WHAT WILL GENETIC TESTING TELL ME

Types of Test Results

Positive

The test found a genetic change that is known to cause the symptoms or a health condition.



Negative

The test did not find a genetic change that is known to cause the symptoms or a health condition.



VUS

Variant of Uncertain Significance: the test found a genetic change, but it's not yet known whether it causes the symptoms or a health condition.



Polygenic Risk Scores (PRS): This test found multiple genetic changes that together show someone might be at a low, average, or high risk for a condition.

VUS* and PRS are NOT diagnostic. Only a positive or negative result can be used to make or rule out a diagnosis. However, it's important to remember that a genetic test will only tell us about what we tested for.

Example: a negative result only means a patient does not have the specific condition(s) tested for. The patient still may have a different genetic condition that was not tested for.

*Genetics research is increasing quickly. With more time and research, a VUS can be recategorized as positive or negative. It's important to keep in touch with a genetics provider. People who are NOT of White/European ancestry are more likely to have a VUS found because there is less research in these groups.