

Understanding a Genetic Test

Not all genetic tests provide the same information. Use these three questions to think through a genetic testing situation: a test you're considering, a test you've already had, or someone else's test.

You don't need to know every detail. Knowing what to ask is part of understanding a genetic test.



1. Why did someone get the test?

Think about what led to testing. Was there a specific health concern, symptoms, or a family history? Was the person curious about their health or ancestry? Was testing offered broadly as screening, or as part of a research study? Or was there another reason?

What I know

What I still need to ask



2. How did they get the test?

How was the test obtained? Consider the four approaches you explored in the module:



Clinical testing



Population screening



Consumer testing



Research testing

More than one approach can apply to the same situation. For example, a research study that offers genetic testing to adults without a specific medical reason is both research testing and population screening.

What I know

What I still need to ask



3. What did the test look for, and how was the DNA examined?

Knowing why someone was tested or how they got the test doesn't tell you exactly which genetic variants the test looked for or could detect. Consider:

Method

How was the DNA analyzed? For example, was genotyping or sequencing used?

What I know

What I still need to ask

Amount of DNA

Did the test look at a specific variant, a group of genes, or nearly the whole genome?

Types of variants

What types of genetic variants was the test designed to detect? For example, SNPs and other small variants, larger deletions and duplications, or both.

These details may be in the test report, or you can ask a healthcare professional, the testing company, or the research team.

Why these questions matter

Understanding why someone was tested, how they got the test, and what the test was designed to look for can help clarify what its results can and can't tell them.

This is especially important for a "negative" result (no genetic variants of interest detected). A test can only identify variants it was designed to look for and detect.