

Next generation sequencing (aka panel testing) is a relatively newer technology that allows the lab to look at many different genes in a faster and more cost-effective way than ever before. This type of testing can be particularly helpful in situations where there are many potential explanations for what we see in a personal or family history.

Before this testing existed, doctors would go through the list of possible genetic conditions that explain the medical and family history one by one starting with the most likely. Test results for each gene would take anywhere from two to 12 weeks to come back, and the cost for testing per gene was anywhere from \$500 to \$5000!

Next generation panel testing can allow the lab to look at several genes at one time, and results are generally back within three to 12 weeks, depending on the type of testing and the lab. The price of panel testing can be up to several thousand dollars, but is on the decline for most types of tests. Given the price and time burden of testing each gene individually, in many cases it may be more time and cost-effective to consider a panel test.

That sounds amazing! Why wouldn't I get panel testing?

While panel testing has shown to have many benefits, there are also some potential drawbacks to consider:

Uncertain Guidelines

Some labs are offering testing for genes, particularly some that are linked to an increased risk for <u>cancer</u>, that have no official guidelines. An example of this would be:

• Ms. Abernathy was seen by her genetic counselor because she has a strong family history of <u>breast cancer</u>. The testing came back that she has a harmful change, or pathogenic variant, in a gene that some research studies show is likely related to an increased risk for hereditary <u>breast cancer</u>. However, not enough data has been gathered to show how high the risk is, or what medical management recommendations should be for women who carry a pathogenic variant in this gene. This can create a difficult situation because we have some information, but not enough to be able to tell someone definitively what to do next.

Variants of Uncertain Significance

Another potential concern with panel testing regards variants of uncertain significance (VUS). Visit our <u>results</u> page to learn more about VUS findings.



Unexpected findings

Sometimes genetic testing can be ordered to look for an answer to a potential health concern, but can provide unexpected, although ultimately helpful, information.

Let's say Ms. Abernathy from above was seen by her genetic counselor for a family history of <u>breast cancer</u>, and her genetic counselor ordered a panel test that looked to see if she has an increased risk for various different types of cancer. Her testing came back negative for all of the genes related to hereditary <u>breast cancer</u>, but they did find a pathogenic variant in a gene related to an increased risk for <u>colon cancer</u>.

So, while the test was not able to provide an explanation for her family history of <u>breast</u> <u>cancer</u>, it does give her information that she is at an increased hereditary risk for <u>colon</u> <u>cancer</u>. Ms. Abernathy and her doctor can use this information to begin <u>colon</u> <u>cancer</u> screening at an earlier age and repeat them more frequently than she would otherwise get them in order to keep her healthy.

Another concern with genetic testing is that it may reveal non-paternity (someone's father is someone different than who they thought), which is thought to occur in 2-10% of all people. While thought to be relatively rare, certain types of genetic testing may bring this to light, potentially causing additional anxiety and stress.

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• Next Generation Sequencing/Panel Testing

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• Traditional Chromosome Analysis

A chromosome analysis, or karyotype, is the traditional testing that can been done on cells from chorionic villus sampling (CVS) and amniocentesis procedures, or from a blood sample. A karyotype involves the lab examining the cells and determining the number of chromosomes. How the lab determines the number of chromosomes...

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A chromosomal microarray (CMA) is a test that can be done to look for specific kinds of chromosome changes called microdeletions and microduplications. Much like a traditional chromosome analysis, CMA generally looks at all 23 pairs of chromosomes. While the chromosome analysis is looking for large changes (extra or missing...

<u>Genetic Testing Results</u>

With genetic testing, there are generally three different results that we can get: a positive, a negative, or a variant of uncertain significance (VUS). Positive A positive result means that a harmful change (called a pathogenic variant) was found in a gene that causes that gene to not work properly....

• How to Decide

Whether or not to pursue any genetic testing is a very personal decision. In some cases, moving forward with genetic testing may help to provide an answer to a health question that is running in someone's family, or may help to provide a name to a medical condition that someone...