A *positive* result (may also be called ‘mutation detected’) confirms inherited risk for cancer in the person who was tested. A mutation means that the lab found either a spelling error (by sequencing), or missing or extra letters (by deletion/duplication studies) that we know make the instructions for that gene incorrect. If the instructions are incorrect, then that gene will either not produce a protein, or will produce a protein that doesn’t do what it is supposed to. In the case of hereditary cancer genes, if they are not producing a protein, or are producing a protein that doesn’t function properly, these non-working genes can cause an increased risk for related types of cancer.

Each gene is related to different types of cancer. Therefore, if someone has a mutation in a gene related to hereditary cancer, identifying which gene the mutation is in can tell us what forms of cancer someone may be at risk for, and how high that risk is. Once we know that someone is at an increased genetic risk for cancer, appropriate high-risk screening and management options should be discussed.

Finding out that someone carries a gene mutation associated with an increased risk for hereditary cancer means that their relatives may also be at an increased risk. Most of these genes are passed down in an autosomal dominant pattern, meaning that if someone carries a gene mutation, there is a 50% risk for first-degree relatives (parents, siblings, and children) to also carry it. If we have an identified familial gene mutation, then we can use that information to offer testing to family members to determine who may also have a higher hereditary risk for cancer. If relatives are also found to carry this gene mutation, then recommendations can be made for high-risk screening and management. If relatives are found to not carry the gene mutation, then we can reassure them that they should have a risk for cancer that is similar to those in the general population.

Click here to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.

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