Genetic testing for hereditary cancer first starts with a genetic consultation to collect and analyze the personal and/or family history of cancer. If a pattern suggestive of hereditary cancer is identified, then genetic testing may be considered for further clarification or confirmation of the specific cancer risks.

Depending on the history, it may be recommended that a specific individual in the family is the first to undergo the genetic testing. Whenever possible, it is recommended to initiate testing in a family member who has had a diagnosis of cancer most suggestive of hereditary causes, since this is the most likely person to have a mutation identified through the testing. However, we understand that this is not always possible, such as when the family member is deceased or is otherwise unable or unwilling to undergo the testing.

If a cancer-associated gene mutation has previously been identified in the family, then testing can be targeted to that specific mutation with the ability to definitively confirm or rule out increased risk. If no prior testing has been done, then genetic testing may include one or a few specific genes that are strongly suspected based on the history. Alternatively, in some cases, more broad panels to include testing of multiple genes may be considered.

To help understand the possible results, it is important to have a brief background about what genetic testing looks for. Each of our genes is made up of a string of thousands of letters. These letters make up an instruction manual for how to build a protein, and the proteins are what is actually making our body function how it should. Genetic testing generally looks at genes through sequencing and deletion/duplication analysis. Most of the genes that we associate with hereditary cancer risk make proteins that help our bodies prevent various types of cancer from forming.

- **My results are positive – what does that mean?**
- **My results are negative – what does that mean?**
- **My results say I have a variant of uncertain significance – what does that mean?**

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

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