sema4

Prior to ordering genetic testing, it is important to make sure that the patient has a good understanding of the test ordered and its implications. This guide is for informational purposes only to provide a better understanding of some key elements of genetic testing and does not replace genetic counseling or other treatment services.

Eliciting the personal and family history

By using the Family History Questionaire or other tools to elicit relevant medical history, the treating physician can obtain the necessary personal and family history information from his/her patient. It is important to ask the patient whether there is already a known genetic change identified in the family. If this is the case, the patient could benefit from a referral to a clinical genetics department for a more comprehensive genetics consultation.

Information about the patient's personal and family history should be included with the test sample.

This information can help the ordering healthcare provider select the most appropriate test for their patient. Additionally, Genetic test results should always be interpreted in the context of a patient's personal and family history. The presence or absence of a significant history can also impact the medical management of the patient.

+ Positive	This means that a pathogenic or likely pathogenic variant that is associated with an increased risk to develop cancers/tumors. The specific type(s) of these risks can vary depending on the gene in which the variant is found. A positive result may have medical management implications for the patient and for family members. For certain genes/variant, the cancer risks are not well established at this time, and more data and specific medical management recommendations related to these genes may evolve over time.
– Negative	This means that no pathogenic or likely pathogenic variants were identified. A negative result reduces, but does not eliminate, the possibility that the patient carries variants in the genes analyzed or in other genes that are not included in the test. The patient may still have the general population's risk for developing cancer and may also be at a higher risk to develop cancer based on personal/family history.
~ Inconclusive	This means that an alteration was identified in the genes tested and there is insufficient information at this time to determine if such alteration is associated with increased cancer risks. A variant of uncertain significance (VUS) is not a positive test result. No medical actions should be taken solely on the basis of a VUS result.

These results can have significant implications for the patient's medical management. By understanding the types of results, a patient can make a more informed decision about whether they want to proceed with genetic testing. Genetic test results may also impact the patient's family members because genetic changes can be inherited.

Limitations of testing

It is important for patients to be aware that there are limitations to genetic testing. One example is that this test does not examine every gene or genetic change associated with cancer predisposition. Therefore, while a negative result may indicate a reduced chance of the patient having hereditary cancer, it does not eliminate this possibility. Other limitations to testing include those discussed in the Sema4 Informed Consent for Hereditary Cancer Genetic Testing.

GINA

There are some federal and state laws that address genetic discrimination. The US Genetic Information Nondiscrimination Act (GINA) may prohibit discrimination by employers and health insurances. This law, however, does not protect people in the military nor possible discrimination by other types of insurance such as life, disability or long-term care.

Some patients may delay testing until they have obtained certain insurance policies, such as life insurance. If such delay is requested by the patient, consider collecting the sample when the patient is ready to continue the testing process. Please go to **http://www.ginahelp.org** to learn more.

What to expect in the post test tele-genetic counseling session?

The genetic counselor will review the patient's personal and family history provided as well as the genetic result. The genetic counselor will discuss the significance of the genetic finding(s) and how it might impact the patient and/or their family members' medical care. Sometimes, the genetic counselor will make appropriate recommendations for additional testing and follow up care with certain medical specialties. The genetic counselor will also discuss family members who may benefit from genetic counseling and testing.