Yale NewHaven Health
Smilow Cancer Hospital

Yale CANCER CENTER
A Comprehensive Cancer Center Designated by the National Cancer Institute

Genetic testing
Information for patients

Purpose of genetic testing
The purpose of genetic testing is to determine whether a person inherited a change, also known as a pathogenic variant or mutation, within gene(s) involved in the development of hereditary cancer syndromes. This testing generally requires a saliva or blood sample.

Limitations of genetic testing
Like most medical tests, genetic testing has limitations. Due to limitations of genetic testing technology, it is possible that a pathogenic variant in these gene(s) may not be detected or may be present in a gene that was not analyzed. Genetic testing does not provide information about a person's current health status.

Psychological consultation
Since this testing has implications for both an individual and their family members, it can be an emotional process for some people. Some people undergoing genetic testing find it helpful to consult with a trained psychiatrist, psychologist or social worker to discuss these issues prior to receiving their results. The Smilow Cancer Genetics and Prevention Program can provide a referral if needed.

Results
Results of genetic testing are disclosed by telephone or at an in-person, follow-up consultation. There are three possible outcomes of this testing:

1. A pathogenic (disease-causing) variant (mutation) is found, which has implications for the person undergoing testing, their children, and their family members.

2. No variants are detected. However, this does not rule out the possibility of a pathogenic variant in these gene(s) that is not detectable by the current testing technology or other cancer susceptibility factors.

3. A variant of uncertain significance (a genetic change whose significance is not yet known) is detected and as a result, the implications may be unclear.

Benefits of genetic testing
Studies show people who receive a positive test result and those who receive a negative test result benefit from this information.

A positive test result (finding a pathogenic variant) can clarify an individual's own cancer risks, as well as risks to their children and other relatives. This may allow them to be better equipped

to plan for the future and have the opportunity to develop a personalized cancer prevention and screening plan.

A true negative test result (not finding a mutation in an individual when the mutation in their family has been previously identified) can offer reassurance and may lead to reduced anxiety since there would be no increased cancer risk to them or their children.
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An uninformative negative test result (not finding a mutation in an individual when the mutation in their family is not yet known) can also offer some reassurance and may lead to some reduction in anxiety as it may somewhat decrease the risks to them and their children.

Risks and discomforts of genetic testing:

There are no unusual risks involved in collecting a saliva sample or having blood drawn. A blood draw may include pain, bruising, and bleeding or lead to an infection at the site of the needle stick (although infection is a rare complication).

There are several psychological and discrimination issues that need to be considered.

Receiving a positive test result may lead to depression, heightened anxiety and fear. A positive test result may also bring about changes in family dynamics or stigmatization by family members or friends. In addition, there may be concerns of discrimination by an employer and/or insurance company (particularly life and disability). However, there are laws, such as the Genetic Information Non-Discrimination Act (GINA), in place that provide some protection from discrimination based on genetic test results, particularly with regard to employment and health insurance.

A true negative test result may cause feelings of guilt. This is often called ”survivor guilt” and is experienced when one member of the family tests negative, while others test positive.

An uninformative negative test result or variant of uncertain significance test result may cause feelings of frustration, confusion or disappointment since the results may not provide definitive information about a person’s risks, risks to other family members, or the cause of the cancers in their family. Uninformative negative or variant of uncertain significance test results may also provide individuals with either a false sense of reassurance or anxiety about their cancer risks.

In the case of gene panel testing performed through the Yale DNA Diagnostics Laboratory, if no detectable mutation is identified in the genes tested, the laboratory may look for other, less common gene mutations associated with cancer. In this case, the laboratory will only report test results that will have a direct impact on the person’s medical care. Testing also has the ability to detect rare pathogenic variants (incidental findings) associated with diseases *other* than cancer. For diseases in which medical intervention is available, such results will be reported to the individual undergoing genetic testing.

Confidentiality

After results disclosure, a summary letter with a copy of the test results will be sent to the person undergoing testing and their referring health care provider. The insurance company of the person undergoing testing may become aware of this information if they are paying for testing or if they are otherwise given permission to access the information.

Information about genetic testing

The Smilow Cancer Genetics and Prevention Program is available to answer any questions that you may have. You may contact the genetic counselors at 203.200,4362 if you have any new questions about testing or the testing process.

Future correspondence

As technology continues to change and new data in the field of genetics emerges, it is likely that there will be additional information about hereditary cancer in the future that may be important to people and their families. We encourage all individuals undergoing genetic testing to keep in contact with the Smilow Cancer Genetics and Prevention Program on a regular basis, particularly if there are any changes in their personal or family history.

Individuals may also find it helpful to check the program's website for updates.

Contact information

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