

Hereditary Cancer and Multi-Gene (Panel) Testing

All cancers are caused by changes (mutations) to genes. Genes are the set of instructions for the cells of the body. When there is a mutation in a gene, this causes it not to work properly. When certain genes are not working properly, this can cause abnormalities in the cell to build up which could lead to cancer.

In most cancers, the gene mutations occur due to many exposures, risk factors and normal aging. However, about 5 to 10 percent (%) of cancers develop due to a mutation in a gene that was present since birth. This is called hereditary cancer or a hereditary cancer syndrome. Gene mutations that cause hereditary cancer syndromes can be passed down in families. Individuals who inherit these gene mutations have an increased risk of developing cancer. Typically, each child of a person with the hereditary cancer syndrome will have a 50/50 chance to inherit the mutation. It is important for individuals with hereditary cancer, as well as their family members, to consider cancer risk-reduction and early detection as directed by their health care providers.

Types of Hereditary Cancer

Types of hereditary cancer include breast, colon, ovarian, uterine, kidney, skin and others. Some hereditary cancer syndromes can increase risk for more than one cancer type. Some genes greatly increase cancer risk, others only moderately increase cancer risk and some are not well studied so the increased risk is not yet known. Genetic testing can look for mutations in one or more genes that are associated with hereditary cancer.

Features of Hereditary Cancer

There are several factors associated with hereditary cancer in a family. These can include:

- Close relatives with breast, ovarian, colon or other cancers.
- Cancer diagnosed at a younger than average age.
- Multiple cancer types in the same individual or family that are part of a specific syndrome.
- Rare cancer types, such as male breast cancer.
- Certain ethnic background, such as Ashkenazi Jewish ancestry.

Further tests may be needed if a person's medical or family history suggests the possibility of hereditary cancer. If there is concern, it is important to discuss personal and/or family history with a health care provider. This may lead to a referral to the Cancer Genetics Program of the MD Anderson Cancer Center at Cooper for genetic counseling and discussion of genetic testing options.

Genetic Testing for Hereditary Cancer

Genetic testing is done using a blood or saliva sample. Genetic testing can look for mutations in the main genes which cause a hereditary cancer type, such as BRCA1 and BRCA2. Laboratories can also test for mutations in many different genes associated with hereditary cancer. These are called "multi-gene panel tests."

Multi-gene panel tests can look for mutations in genes that cause a specific type, such as breast cancer, or multiple types of hereditary cancer.

Multi-gene panel tests can look for mutations in genes known to cause a very high risk for cancer. For many of these genes there are national guidelines to help doctors and patients make decisions about managing cancer risk.

Some multi-gene panel tests also look for mutations in genes that moderately increase cancer risk and/or in those in which the exact cancer risk is not yet known. These genes have not yet been well studied. In these cases, research is still ongoing and there may not be specific national guidelines to help make decisions about managing risk.

Candidates for Panel Testing

Depending on the situation, testing for only one or a few genes may be most appropriate. However, multi-gene panel testing might be an option for individuals with any of the following:

- Negative or uncertain genetic test results in the past.
- Personal and/or family history of cancer that is suggestive of more than one hereditary cancer type.

Possible Results of Panel Testing

There are three possible outcomes of panel testing. These include:

- Positive for a mutation. This confirms a diagnosis of hereditary cancer. Other family members can have genetic testing to learn whether or not they have the mutation and the hereditary cancer syndrome.
- Negative for a mutation. Cancer is not likely related to a mutation in the genes tested. However, a hereditary cause cannot be completely eliminated as there could be a mutation not detectable or a mutation in a gene that was not tested.
- Variant of Uncertain Significance. A gene change that does not provide clear information about cancer risk. An uncertain result means that we do not yet know if the gene change detected in an individual increases risk for cancer or if it is benign (harmless) variation. Until researchers learn more about whether the change leads to increased risk, medical decisions cannot be made based on the result.

Potential Benefits of Panel Testing

Some of the benefits of panel testing include:

- Explain why cancer developed in the family.
- Guide medical decisions about cancer risk management and reduction.
- Provide information about cancer risk for family members.

Potential Risks or Limitations of Panel Testing

Some of the potential risks or limitations of panel testing include:

- Negative results cannot completely eliminate the possibility of hereditary cancer.
- Uncertain or inconclusive results could cause worry or concern.
- Unexpected positive results could diagnose a hereditary cancer syndrome that would not have been anticipated based on the personal or family history.
- National guidelines for risk management may not be available for all positive results.

If you are concerned about the possibility of a hereditary cancer syndrome in your family, you are encouraged to discuss your personal and/or family history with your health care provider. Your physician may refer you to the Cancer Genetics Program of the MD Anderson Cancer Center at Cooper for genetic evaluation and discussion of testing options. If genetic testing is warranted and you choose to proceed, a blood or saliva sample will be taken during your visit to start the testing process. Please note health insurance companies may cover most, if not all, of the cost of genetic testing on a case-by-case basis.

Resources

National Cancer Institute

www.cancer.gov

This site has cancer-related information on more than 200 cancer types, clinical trials, cancer statistics, prevention, screening, risk factors, genetics and support resources. Information is available in Spanish. See the “Genetic testing for Hereditary Cancer Syndromes” page.

American Cancer Society

www.cancer.org

The ACS is a voluntary national health organization that supports research, provides information and offers programs and services to patients and their families. Information is available in Spanish. See the “Genetics and Cancer” page.

Genetic Alliance, Inc.

www.geneticalliance.org

This organization supports individuals with genetic conditions and their families, educates the public and advocates for consumer-informed public policies. See the “Publications” page for brochures and guides.

FORCE (Facing Our Risk of Cancer Empowered)

www.facingourrisk.org

FORCE is a nonprofit organization for individuals and their family members who may be at high risk of developing breast and/or ovarian cancer.

Bright Pink

www.brightpink.org

Bright Pink is a nonprofit organization, geared toward young women between the ages of 18 and 45, who are at increased risk of developing breast and/or ovarian cancer.

Gilda’s Club of South Jersey

www.gildasclubsouthjersey.org

Gilda’s Club South Jersey (GCSJ) is a cancer support community in southern New Jersey offering a free, comprehensive program of psychosocial support including educational lectures, workshops, social activities and support groups to men, women, teens and children impacted by cancer.