Somatic and Germline Cancer Testing

Pre-Test Genetic Counseling Information

Somatic testing is done on cancer cells. It can help doctors learn more about your cancer.

Germline testing is done on non-cancer cells. It can show if a genetic change is in all of your cells in your body.

What is genetic testing?

Genetic testing looks for specific changes (mutations) in a person's genetic information. "Genes," "DNA," and "chromosomes" are all genetic information. Genetic information tells a person's body how to grow and function. All people have genetic changes, which makes everyone unique. Some changes can affect health and development.

What is cancer genetic testing?

Cancer genetic testing looks for mutations in cells with cancer (somatic) and cells without cancer (germline). Genetic mutations can have different effects on your health:

- Harmful (can make you sick or cause health problems)
- Beneficial (can be helpful or protective from health problems)
- Neutral (no effect)
- Uncertain effects

Genetic mutations in cells with cancer (somatic) can sometimes give doctors information about how to treat the cancer. Mutations that are identified in cells without cancer (germline) may be associated with health problems passed down from a parent (inherited). These can make someone more likely to have cancer and other health risks. Overall, inherited mutations are thought to play a role in about 5% to 10% of all cancers.

What are somatic mutations?

Somatic mutations are changes that happen to the genetic information in cells after a person is conceived. They are not passed down from a parent. They can happen to almost any cell of the body. The mutations can cause cancer, but not always.

What is somatic testing?

Somatic testing is usually done after a person has been diagnosed with cancer. It looks for genetic mutations in tumor or cancer cells. For some types of cancer, it can help doctors find out more about the cancer, including:

- Information on an exact diagnosis
- Information about the outlook (prognosis) of the cancer
- If treatments are available for cancer with that specific mutation

Usually mutations in cancer cells are only in those cells. They are not in the rest of the cells of the body. Somatic genetic tests look for gene changes only

in cancer cells. These tests are not intended to give information about cancer risk passed down from a parent (inherited).

What are germline mutations?

Germline mutations are a genetic change in all of the cells in your body. These changes usually happen before conception and were usually, but not always, passed down from a parent.

What is germline testing?

Germline testing is done on cells that do not have cancer. It is done to see if a person has a gene mutation that is known to increase the risk of developing cancers and other health problems. This test uses cells (such as blood or skin cells) that do not have any cancer cells. Germline mutations can sometimes be passed down from parents.

Your healthcare team may recommend germline testing for you or your child if cancer cell (somatic) testing finds a mutation. This helps your doctor see if the mutation is in all of the cells of the body, or just the cancer cells.

What kind of cancer genetic tests are there?

Chromosome tests

Chromosome tests look for extra or missing pieces of chromosomes. Chromosomes are packages of genes or DNA. Finding extra or missing copies of genetic material can sometimes help a doctor diagnose and treat the cancer. A cancer microarray and karyotype are 2 types of chromosome tests. They can't find all differences in a person's genetic information.

Gene tests

Gene tests, also called molecular tests, look for mutations in genes. Genes are small pieces of genetic information found in chromosomes.

What do the germline test results mean?

Normal/Negative: This result means that the test did not find any changes in the tested genes. This could mean that you are:

- Not affected by the disorder
- Do not have an increased risk of developing the disorder
- Are not a carrier of the specific genetic mutation

However, 1 kind of test cannot look for every possible genetic change. You may need more tests.

Abnormal/positive: This result means that a change was found in the tested genes. This usually will:

- Diagnose a genetic disorder
- Confirm that the person is a carrier for a disorder
- Identify an increased risk of developing a disease

To Learn More

- Ask your child's oncology provider
- See our handout "Medical Genetics" www.seattlechildrens.org/pdf/PE2386.pdf

Free Interpreter Services

- In the hospital, ask your child's nurse.
- From outside the hospital, call the toll-free Family Interpreting Line 1-866-583-1527. Tell the interpreter the name or extension you need.

A *_de novo_* mutation, also called a new mutation, is a genetic change that happens for the first time in only 1 family member. A *de novo* mutation may explain genetic disorders when there is no family history of the disorder and the child with cancer has a mutation in every cell in their body, but the parents do not have the same mutation.

Variant of uncertain significance (VUS): This result means that the test found a genetic change, but there is not enough known about the change to give a diagnosis. In some cases, testing other family members can help us understand what the genetic change means. It is more common to find this test result when many genes are tested at once (panel test). Finding a VUS does not typically change medical management recommendations. Your doctor will make recommendations based on personal and family history of cancer until more is learned about the VUS.

Unexpected/incidental result: A genetic test may find an important change that is not directly related to the reason for testing.

Other results: Genetic testing can also provide information on the biological relationships in a family. Examples of this include:

- Finding that the stated father of an individual is not the biologic father
- Finding that the parents may be biologically related to each other

Does a somatic test result mean the cancer is inherited?

No. Mutations that are found in cancer cells are only sometimes (or rarely) also found in the person's germline cells. Testing another sample without cancer, such as cells from your blood or skin, may be needed to see if a mutation is inherited.

Insurance pre-authorization

Genetic tests are often expensive and insurance may not pay for them. You may have to pay for the test yourself if the test is not pre-authorized. To learn more, read our handout "Insurance Coverage for Genetic Testing" www.seattlechildrens.org/pdf/PE2051.pdf.

Genetic Counseling

A genetic counselor is available to talk about cancer genetic testing with you. Ask your provider for more information about a referral to a genetic counselor.

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