ARIZONA BLOOD & CANCER SPECIALISTS

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UNDERSTANDING GENETIC TESTING

Every person has their own unique set of genes that are passed down from their parents at birth. A person's genes can determine their physical features such as hair color, eye color, and height but they can also affect whether a person is likely to develop certain disorders or diseases.

Genetic tests are done to look for specific changes or mutations in a person's chromosomes, genes or proteins. There are many types of genetic tests including those to detect genetic changes or mutations known to cause cancer.

The different types of tests are categorized as follows:

Predictive Test
A predictive test indicates whether a person has an increased chance of developing a disease or condition. This type of test cannot predict with certainty whether a person will develop the condition, or when symptoms will appear.

Pre-symptomatic Test
A genetic test is considered a pre-symptomatic test if the result indicates that a person will eventually develop a disease or condition, however this type of test cannot tell exactly when the disease or condition will occur.

Diagnostic Test
While a diagnostic genetic test can confirm a suspected diagnosis, it may not give specific information about how severe a person's disease or condition currently is or will be in the future.

Genetic Mutations and Cancer

Cancer is caused by genetic mutations, and sometimes these genetic mutations are hereditary. When a person's genes have certain abnormalities or mutations, there is a greater risk of developing cancer. Mutations can affect how cells grow and divide. Sometimes mutations can cause cells to grow out of control which can lead to cancer.

For people with a family history of cancer, genetic testing is an important tool that can help identify the potential for problems, make decisions to monitor for early detection, or to decrease or prevent the chance of a future disease. It is important to note however, that only about 5–10% of cancers are caused by an inherited genetic mutation.

Cancer can be a result of acquired gene mutations which are mutations that happen during a person's lifetime. Sometimes these mutations can be from a known cause; for example, from smoking / tobacco use or from the sun's harmful UVA / UVB rays. Other times the genetic mutations can occur without any clear cause.

Some people think that "cancer runs in their family," but the cancer may be a result of a environmental exposure or a similar behavior such as having several members of the same family that smoke.

Understanding your family's medical history

Doctors typically ask about your first-degree relatives who include your parents, siblings, and children, but now we are finding that it is also important to know about your second-degree relatives too. These would be your grandparents, grandchildren, uncles, aunts, nephews, nieces, and half-siblings.

Gathering this data can help identify whether members of the same family have cancer, or other common inherited disorders that include obesity, heart disease, high blood pressure, arthritis, diabetes, and Alzheimer's disease.

It also can provide important information to your doctor about certain genetic mutations that may be present among several family members. In some families, many relatives on one side of a person's family may all have the same type of cancer. There may also be different types of cancer that are all linked to the same genetic mutation.

Other clues that doctors look for is if a family member has had more than one type of cancer, family members who have been diagnosed younger than the typical age for certain types of cancer, or family members who have been diagnosed with rare types of cancer.

A person's ethnicity can also be an indicator that genetic testing may be needed. Specific ancestries are often linked to a higher prevalence of certain types of cancer.

Physical findings discovered during cancer screening (for example, colon polyps) are also linked to certain inherited cancers.

Based on your family history, your doctor can make a recommendation for genetic testing to better understand if you are at a higher risk, determine if further testing is necessary, customize a cancer screening plan, and suggest strategies that can help lower your risk for developing cancer.

For patients who already have been diagnosed with cancer, genetic tests can help their oncologist identify the most effective treatment options.

The Centers for Disease Control has an online tool from the Surgeon General to help you document your family's medical history. "My Family Health Portrait" can be shared with your family or your health care provider.

Can I use a home-based family ancestry kit to learn about my genes?

While it can be fun to learn about your ancestry, it's also important to know your family's medical history. The popular tests that can be ordered online do not provide enough information to determine a person's overall risk of cancer or other diseases.

Should I be tested?

Just as every cancer is different, so is each person's situation. While some people may choose to have testing, others decide not to. Genetic testing is completely voluntary, and you are the only person who can make the decision that is right for you.

We recommend that you discuss any questions you have with your doctor. It is also important for you to meet with a genetic counselor prior to being tested (www.nsgc.org/page/find-a-genetic-counselor). They can help you with knowing what to expect. They will help you explore the pros and cons of being tested and can help you understand what the results mean as well as what options you have.

Will insurance cover the cost of genetic testing?

When genetic counseling and testing is ordered by a doctor, health insurance policies will typically cover the cost, however it is important for patients to check with their insurance company to verify coverage for these services prior to being tested.

Sometimes, genetic counseling and testing is covered only under specific circumstances, or sometimes there are stipulations where certain requirements need to be met before the counseling and testing are covered.

Risks of testing

Typically, there is minimal physical risk from genetic testing. The test is usually done with a sample of a person's blood. Sometimes tissue samples may be necessary. The procedure used to collect the sample should be explained in detail, so that you are aware of any associated risks.

There is a potential for both psychological and social risks of having genetic testing. The decision to have the testing can be stressful while anticipating what the results will be, or how your life may be affected. Genetic testing can also have an impact on family relationships for a variety of reasons.

Like any other medical test or procedure, genetic tests do become part of your permanent medical record. A person's medical record is strictly confidential and protected under the Health Insurance Portability and Accountability Act of 1996 (HIPAA), a federal law to protect sensitive patient health information from being disclosed without the patient's consent or knowledge. While laws are also in place to prohibit discrimination, people are often fearful that the results of their tests can negatively reflect on them in the future by future employers or other health insurers.

Accuracy of genetic tests

Genetic testing is highly accurate, but there are some limitations, and sometimes do not provide a definitive answer. Your doctor will review your test results with you and answer any questions you may have. Together, you can make decisions about the next steps including any follow up testing and actions you can take to lower your risk including cancer screening, and lifestyle choices that can decrease or prevent the chance of future disease.

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ATENCIÓN: si habla español, tiene a su disposición servicios gratuitos de asistencia lingüística. Llame al: 1-800-201-7220.

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