Great Valley OB/GYN
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Genetic Testing for Cancer Prevention: from the desk of Carol Coldren, M.D.

Genes are responsible for inherited traits such as eye and hair color that are passed on from parents to children. They also affect a person's risk of developing certain diseases, including cancer. Mutations in the genes play a role in the development of cancer by altering the production of the proteins that control cell growth, division and death. For instance, some mutations cause cell division to accelerate - one of the mechanisms that can lead to the development of cancer. Most mutations are not inherited and are either random events or are triggered by environmental exposures to carcinogens including tobacco and radiation. However, if there is a pattern of disease within a family and/or family members have developed a disease at a younger age than usual, there is a higher likelihood that the disease is related to a genetic mutation. Some breast, ovarian and colon cancers, among others, have been linked to single gene mutations, allowing for genetic testing. For instance, testing for mutations in the tumor suppressor genes BRCA1 and BRCA2 is available. People with either of these mutations have a greatly increased risk of breast cancer as well as ovarian cancer for women and prostate cancer for men. Hundreds of different types of mutations have been identified in these genes, some harmful and others seemingly benign or unknown. Only 5%-10% of breast cancers in women have been linked to an inherited gene mutation. In other words, the vast majority of women diagnosed with breast cancer do not have a known inherited gene mutation.

Consequently, while testing is available for identifying BRCA mutations, it's generally only recommended for certain people such as those with a known BRCA1 or BRCA2 gene mutation in a close relative (a parent, sibling, child, grandparent, grandchild, aunt, uncle, niece or nephew), a personal history of breast cancer at 40 or younger, or bilateral breast cancers at any age, a personal history of ovarian, fallopian tube or peritoneal cancer, a family history of breast cancer in two or more close relatives. It is generally recommended that this type of genetic testing be deferred until after age 21. Positive test results may provide incentive for the testing of additional family members. In most cases, initial testing is performed on the person diagnosed with breast or ovarian cancer. If a BRCA1 or 2 mutation is found, other family members may be eligible for testing. There are three possible results from the testing: no mutation, a mutation linked to cancer (positive or carrier) or a mutation not currently known to increase breast cancer risk (called a variant of uncertain significance).Genetic testing for gene mutations provides a better estimate of an individual's risk of developing the disease. Being a carrier of a gene mutation known to increase the risk of cancer does not mean that type of cancer will definitely develop. Likewise, not being a carrier of known risky gene mutations does not guarantee that type of cancer will not develop.

The decision to pursue genetic testing is a very personal one and genetic counseling can be very helpful. The prospect of learning that one is a carrier of a gene mutation that can cause cancer can be frightening. Family relationships may be affected, should family secrets such as paternity, adoptions, or other difficult issues need to be revealed. In some cases the results may generate the need for additional medical tests or procedures which can cause even more stress and anxiety. There may also be concern that a positive result will cause difficulty in obtaining medical insurance, or workplace discrimination, even though the Genetic Information Nondiscrimination Act (GINA) law makes it illegal for most employers to do this. However, a negative result can help relieve anxiety generated from thinking that the risk to oneself and one's children is greater than the risk in the general population. For some people, learning their results, even if they are positive, brings a sense of empowerment and is less stressful than the unknown. A positive result provides an opportunity to change life style habits known to increase or decrease disease risks. It allows for increased surveillance so disease may be diagnosed earlier. It can help identify women who would benefit from risk reducing medications such as Tamoxifen or prophylactic mastectomy for breast cancer, and/or the surgical removal of their ovaries once child-bearing has been completed.

For most cancers, mutations linked to increased risk have not yet been identified. For others, genetic tests are not yet available even though specific mutations are known to increase risk. Continued research in the field will no doubt increase the understanding of our genetic make-up and allow genetic testing to play an ever larger role in cancer risk assessment, detection, and individualized treatment available in the future.

This article is provided as informational and is not intended to be a substitute for individualized professional medical advice.