

Genetics and Breast Cancer

What are genes?

Genetic material or genes are in every cell of your body. Each gene is made up of a unique sequence. Genes code for proteins which can act as messengers in the cell and proteins are the building blocks for the human body. It is important that genes have the proper instructions for making the protein so that the protein can perform correctly. However, genes do not always function correctly, which can be due to variations in the DNA sequences known as mutations.

Mutations can be either inherited or spontaneous. Inherited mutations are those you are born with, while spontaneous mutations happen after a person is born. There are many ways spontaneous mutations can occur, yet it is not known if these mutations are the result of lifestyle, chemical changes in the body or contact with environmental toxins.

BRCA Genes and Breast Cancer

Two genes, BRCA1 and BRCA2, which stand for BREast CAncer gene 1 and 2, have been discovered to be important in hereditary breast and ovarian cancer. Normal BRCA1 and BRCA2 are tumor suppressor genes—suppressing tumor growth. When they are mutated they are no longer able to suppress abnormal growth, which can increase the risk for cancer to develop. Everyone has these genes, but a small number of individuals may have inherited a mutation in one of these genes that increases their risk for breast and ovarian cancers. Having a mutation in BRCA1 or BRCA2 increases the risk for breast and ovarian cancer but doesn't guarantee a person will get cancer.

Approximately, 5% of women with breast cancer will have a mutation in BRCA1 or BRCA2. About 10% of women with ovarian cancer will have a mutation in one of these genes. Women with a family history of breast cancer and/or ovarian cancer, early onset breast cancer or multiple primary cancers are more likely to have an alteration in BRCA1 or BRCA2. Most cancer is caused by a combination of hereditary and environmental factors.

BRCA1 and BRCA2 are just two genes known to cause hereditary cancer. There are hundreds of genes known to be responsible for breast and other cancers.

What is Genetic Testing?

Genetic testing is the analysis of genes, chromosomes and proteins. Genetic testing can identify inherited mutations that increase the risk to develop cancer. Different tests are offered to individuals depending on the spectrum of cancer in their family. The testing is offered first to an affected individual who is likely to have the inherited mutation causing the cancer in the family. If a mutation is identified, predictive testing can be offered to unaffected family members to help them better understand their risk to develop cancer. No genetic test can report with 100% certainty that a person will develop cancer and testing cannot identify all hereditary predispositions to cancer. However, because mutations increase risk, knowing one's genetic status may allow an individual to take

steps to decrease the occurrence of breast cancer or other cancers or detect it in the early stages.

There are both risks and benefits that should be considered before genetic testing is done, such as physical, emotional and financial impact of knowing one's genetic status. Therefore, testing is generally offered only when there is a reasonable likelihood of identifying a genetic change, and the test results will impact the individual's medical care.

Genetic testing can help a person and family understand both the cause and risk of developing cancer. Testing can help in developing a care and cancer prevention plan such as avoiding risk factors, making lifestyle changes or taking preventive medication. If a familial mutation is identified in the family, individuals can find out whether or not they have the cancer risk in the family—if negative, this can be a relief. However, negative testing for familial mutation doesn't eliminate all risk for cancer. If an individual tests positive, it may cause anxiety, or depression about the possibility of developing cancer. Additionally, genetic testing and counseling can be expensive if it is not covered by insurance.

Who Should Consider Being Tested?

Individuals who may want to meet with a genetics counselor include a person with a family history of:

- Multiple relatives on the same side of the family with the same cancer or cluster of cancers
- Cancer that occurs at a young age (colon cancer below age 50; breast cancer below age 50)
- More than one primary cancer
- Rare cancers (male breast cancer, diffuse gastric cancer, lobular breast cancer)
- A known cancer predisposition gene in the family

Meeting With a Genetic Counselor

A genetic counselor is a health professional trained in medical genetics and counseling. Genetic counselors have a Master's degree in genetic counseling, from a board certified genetic counseling program. Genetic counselors are certified through the American Board of Genetic Counseling.

Prior to a genetic counseling appointment, information about the family history of cancer will be asked, such as type of cancer, age at diagnosis, cancer screening, surgical histories for each family member. For family members with cancer, surgical and pathology reports will often be sought. You may want to consider taking a friend or family member with you to the appointment because a lot of information will be covered and it may help to have another person to take notes and think of questions.

Genetic counselors will ask about your medical history and your family's medical history and evaluate the likelihood of hereditary cancer in your family. The counselor will put

together your family tree, which will include at least three generations, and document who had cancer, the type of cancer and their age at diagnosis. They will provide you with genetic testing and screening options, and help with making decisions about future testing. They may also offer information about diagnosis and treatment options, limitations of the testing procedures, test accuracy, screening and preventive measures. In addition, they will advise you on the privacy and confidentiality of genetic information and the emotional, psychological and social consequences of knowing the test results. After your visit, the genetic counselor will write a summary of your visit and usually a copy will be sent to you and your doctor. Additionally, the genetic counselor may provide you with other information relevant to your family history. If you decide to have testing done the counselor will work with you and the testing laboratory to find out if the test is covered by your insurance company. Once testing is completed your counselor will review the results with you and act as a resource for both you and your family.

What is the Cost?

Cost for testing varies. For each gene that is tested, it costs somewhere in the neighborhood of \$1,000; many tests cost more.

Are Genetic Tests Covered By Insurance?

Most health insurance companies will pay for genetic testing, particularly if the test results will impact medical care. However, every insurance plan varies with what they will cover. Therefore, it is important to check with your insurance plan prior to testing. The genetics counselor can work with you and the testing laboratory to find out if the test is covered by your insurance company.

Is Genetic Discrimination Prohibited by Law?

In Washington State, individuals employed at companies with more than 50 individuals, are protected from health insurance discrimination while they remain on their employers group health insurance plan. Most states, including Washington State, offer health insurance to individuals that are self-employed or working for small employers. However, insurance premiums may not be as affordable as group plans.

In general, individuals who are seeking insurance must disclose their family history of cancer which can put them at increased risk for higher premiums. Few insurers specifically ask about genetic status. Family history of cancer alone can make it hard for an individual to purchase individual insurance plans; genetic testing will not change this, unless the person tests negative for a family mutation.

HIPAA provides some protection for individuals with employer-based health insurance and prohibits insurance companies from using genetic information to deny coverage if a person has not already been diagnosed with cancer. However, there are areas that HIPAA does not cover. For example HIPAA does not prohibit the use of genetic information to charge more for health insurance. It does not limit the collection of genetic information by insurance companies, or prohibit them from requesting that an individual have a genetic test. HIPAA does not limit the release of genetic information by insurers. Lastly,

HIPPA does not apply to individual health insurers unless covered by the portability provision. For a complete description visit the National Human Genome Research Institute website at www.genome.gov.

In 2000, the Department of Health and Human Services released the HIPAA National Standards to Protect Patients' Personal Medical Records. This covers medical records kept by healthcare providers, health plans and health care clearing houses. These standards are not specific to genetic testing, but they do protect the privacy of health information.

The American with Disabilities Act protects individuals with disabilities (defined by the law) from discrimination in the workplace. Some individuals with cancer will meet the criteria for disability. Otherwise, there is limited protection for discrimination in employment and for life insurance. The ADA doesn't address genetic predisposition to disease; it pertains to individuals already diagnosed with cancer or genetic disease that meets the definition of disability.

In many states, legislation has been passed to prevent discrimination; however these protections are not universal. The National Human Genome Research Institute (NHGRI) states that 32 states have legislation about genetic information and the workplace, and 41 states have laws about genetic information and health insurance. In 2000, an Executive Order was issued that prohibits genetic discrimination in the workplace for federal employees.

Nationally, Congress has considered numerous bills to protect individuals from genetic discrimination. In April 2007 the House passed the Genetic Information Nondiscrimination Act of 2007. This act, "...prohibits the improper use of genetic information in health insurance and employment...prohibits group health plans and health insurers from denying coverage to a healthy individual or charging that person higher premiums based solely on a genetic predisposition to developing a disease in the future." To read the complete policy or for current information visit the National Human Genome Research Institute website at www.genome.gov.

Resources

Websites:

American Cancer Society

1-800-ACS-2345

1-866-228-4327 (TTY)

www.cancer.org

Division of Medical Genetics at the University of Washington

206-598-4030

www.depts.washington.edu

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MOST Survivor's Notebook

GeneTests

www.genetests.org

National Cancer Institute

1-800-4-CANCER

www.cancer.gov

National Society of Genetic Counselors, Inc

www.nsgc.org

People Living With Cancer

American Society of Clinical Oncology

1900 Duke Street, Suite 200, Alexandria, VA 22314

Attn: Communications and Patient Information Department

Email: contactus@plwc.org

Phone: 703-519-2927 or 888-651-3038

Fax: 703-299-1014

www.plwc.org

Susan G. Komen for the Cure

5005 LBJ Freeway, Suite 250

Dallas, TX 75244

Phone: 972-855-1600

Fax: 972-855-1605

Helpline: 1-800 I'M AWARE (1-800-462-9273)

www.cms.komen.org

Referrals:

Breast and Ovarian Cancer Prevention Program

Gastrointestinal Cancer Prevention Program

Cancer Prevention Clinic

Seattle Cancer Care Alliance (SCCA)

825 Eastlake Ave E, E2-102

Seattle, WA 98109-1023

Phone: 206-288-1024

www.seattlecca.org

Cancer Genetics Clinic

Division of Medical Genetics at the University of Washington Medical Center

Physician or self-referrals are appropriate. However, often insurance companies require a physician referral. For more information or to set up an appointment call **206-598-4030**.