The Genetics of Breast Cancer

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What are genes?

Genes carry information in the form of DNA within each cell of the human body. Researchers estimate that there are 30,000 different genes in each cell. Genes are packaged onto chromosomes. There are 23 pairs of chromosomes in each cell. One chromosome of each pair is inherited from the person's father and one from the person's mother.

Genes control how a cell functions, including how quickly it grows, how often it divides, and how long it lives. To control these functions, genes produce proteins that perform specific tasks and act as messengers for the cell. Therefore, it is essential that each gene have the correct instructions or "code" for making its protein so that the protein can perform the proper function for the cell.

What role do genes play in breast cancer?

Many cancers begin when one or more genes in a cell are mutated (changed), creating an abnormal protein or no protein at all. The information provided by an abnormal protein is different from that of a normal protein, which can cause cells to multiply uncontrollably and become cancerous.

A person may either be born with a genetic mutation in all of their cells (germline mutation) or acquire a genetic mutation in a single cell during his or her lifetime. An acquired mutation is passed on to all cells that develop from that single cell (called a somatic mutation). Somatic mutations can sometimes be caused by environmental factors, such as cigarette smoke. Most breast cancers (about 90% to 95%) are considered sporadic, meaning that the damage to the genes occurs by chance after a person is born and there is no risk of passing on the gene to a person's children. Inherited breast cancers are less common (5% to 10%) and occur when gene mutations are passed within a family from one generation to the next.

What are the chances a mutated gene is inherited?

Every cell usually has two copies of each gene: one inherited from a person's mother and one inherited from a person's father. Breast cancer usually follows an autosomal dominant inheritance pattern, in which a mutation needs to happen in only one copy of the gene for the person to have an increased risk of getting the disease. This means that a parent with a gene mutation may pass on a copy of the normal gene or a copy of the gene with a mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a gene mutation also has a 50% chance of having the same mutation.

What is a person's average risk for breast cancer?

A woman with an average risk of breast cancer has about a 12% chance of developing breast cancer. Breast cancer in men is rare; out of 1,000 men, one will develop breast cancer.

How can a person know if he or she has inherited a genetic mutation that increases his or her risk of breast cancer?

Only genetic testing [2] can determine whether a person has a genetic mutation. Most experts strongly recommend that people considering genetic testing first talk with a genetic counselor [3]. Genetic counselors are trained to explain the risks and benefits of genetic testing.

How does a person know if breast cancer runs in the family?

Breast cancer may run in the family if first-degree relatives (mothers, sisters, brothers, children) or many close relatives (first-degree relatives, grandmothers, aunts, nieces, granddaughters, cousins) have been diagnosed with breast cancer, especially before age 50.

What is a person's risk if breast cancer runs in the family?

If a woman's first-degree relative developed breast cancer, the woman's risk is double the average woman's risk. If two first-degree relatives developed breast cancer, the woman's risk is five times the average risk. It is uncertain how much a woman's risk of breast cancer is increased when a man in the family has breast cancer. One out of five men who develop breast cancer has a family history of the disease.

Which inherited genetic mutations raise the risk of breast cancer?

There are several genes linked to an increased risk of breast cancer. Some of the most common hereditary cancer syndromes associated with breast cancer risk are described below.

Hereditary breast and ovarian cancer (HBOC) syndrome [4]**.** HBOC is associated with mutations in the *BRCA1* and/or *BRCA2* (BRCA stands for BReast CAncer). Women with HBOC have an increased risk of breast cancer and ovarian cancer [5]. Men with HBOC have an increased risk of breast cancer [6] and prostate cancer [7].

Ataxia telangiectasia (A-T) [8]**.** A-T is a rare disorder associated with a specific genetic mutation. It causes progressive neurological problems that lead to difficulty walking, slurred speech, and difficulty with writing and other tasks. People with A-T have an increased risk of leukemia and lymphoma, and possibly melanoma [9], sarcoma [10], breast cancer, ovarian cancer [5], and stomach cancer [11].

Li-Fraumeni syndrome (LFS) [12]**.** LFS is a rare condition associated with a specific genetic mutation. People with LFS have a higher risk of developing osteosarcoma [13] (a type of bone cancer), soft tissue sarcoma [10], leukemia [14], breast cancer, brain cancer [15], and adrenal cortical tumors [16].

Cowden syndrome (CS) [17]**.** CS is a rare genetic condition caused by a specific genetic mutation. People with CS have an increased risk of developing breast cancer and noncancerous breast changes and noncancerous and cancerous tumors of the thyroid [18] and endometrium [19] (lining of the uterus).

Peutz-Jeghers syndrome (PJS) [20]**.** PJS is caused by a specific genetic mutation and is associated with multiple polyps in the digestive tract that become noncancerous tumors, increased pigmentation (dark spots on the skin) on the face and hands, and an increased risk of colorectal cancer [21], breast cancer, uterine cancer [19], ovarian cancer [5], and lung cancer [22].

Other genes. Other genes may cause hereditary breast cancer. However, more research is needed to understand how gene mutations can increase breast cancer risk and to find other genes that may increase a person's risk of breast cancer.

What is your risk level?

In addition to family history, other environmental and lifestyle factors may increase your risk of breast cancer. Discussing your family history and personal risk factors with a doctor helps you better understand your risk. People with a higher than average risk may benefit from genetic counseling and early detection strategies.

A risk factor [23] is anything that increases a person's risk of developing cancer. Having a particular genetic mutation linked to breast cancer cannot predict that a person will develop cancer. Controllable risk factors, such as eating a balanced diet, maintaining a healthy weight, exercising, limiting alcoholic beverages, and avoiding tobacco products also play a role. Most people who develop breast cancer have few known risk factors. Research to better understand the link between genetic mutations and breast cancer is ongoing. Talk with a doctor for more information about risk factors, prevention, and screening for breast cancer.

More Information

Genetics [24]

Guide to Breast Cancer [6]

Chemoprevention [25]

Sharing Genetic Test Results With Your Family [26]

Direct-to-Consumer Genetic Testing [27]

Links:
[1] http://www.cancer.net/about-us
[2] http://www.cancer.net/node/24895
[3] http://www.cancer.net/node/24907
[4] http://www.cancer.net/node/18922
[5] http://www.cancer.net/node/19481
[6] http://www.cancer.net/node/18618
[7] http://www.cancer.net/node/19562
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[19] http://www.cancer.net/node/19308
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