Genetics of Breast Cancer

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Breast cancer involves uncontrolled growth of cells in the breasts, which is caused by a mutation in genes.

These gene mutations may be inherited from the individual's parents or acquired throughout their lifetime.

The cells in the body are continually being renewed through a process of cell replication and death.

DNA provides the specific information about the cells so that they can be replicated and continue to grow. If there is a small error in the DNA of the cell, that cell will continue to replicate the error further.

In many cases, a single error will not have a significant effect on the function of the cell, but with time successive errors may interfere with the growth or function of the cell, leading to abnormal behavior of the cell.

For example, cancer cells exhibit uncontrolled growth in the body as a result of the genetic mutation, which can take over the health tissue and cause significant health implications.

There are several specific genes that have been linked to an increased risk of developing breast cancer. They will be discussed in more detail below.

BRCA1 and BRCA2 Genes

The most common types of inherited breast cancer genes are BReast CAncer gene one (BRCA1) and BReast CAncer gene two (BRCA2).

All individuals have these genes, which are usually responsible to repair damage to cells in the breast and promote healthy growth of the cells.

However, if an individual has a mutation in one of these genes that interferes with its function, their risk of developing breast cancer significantly increases.

Approximately 1 in 10 of all cases of breast cancer may be attributed to an abnormality in the BRCA1 or BRCA2 genes.

Associated Risk of Breast Cancer

In the United States, each woman has approximately 12% risk of developing breast cancer in her lifetime. However, this risk increases dramatically up to 80% for women who have a mutation in the BRCA1 or BRCA2 gene.

Breast cancers linked to these gene abnormalities tend to present in women at a younger age and affect both sides of the body, in comparison to cases of breast cancer without the genetic mutation.

This increased risk of breast cancer for individuals with the gene mutation holds true for males.

Men with a mutation in the BRCA1 or BRCA2 gene have a risk of 8% of developing breast cancer by the age of 80, which is approximately 80 times higher than for males without the mutation.

The risk of related cancers, such as ovarian, colon, and pancreatic cancer also increases for women with a mutation in one of these genes.

Other Genes Linked to Breast Cancer

In addition to the BRCA1 and BRCA2 genes, there are several other genes that have been associated with an increased risk of developing breast cancer. These genes include:

- ATM gene: helps to repair damaged DNA; mutation associated with an increased rate of breast and pancreatic cancer.
- BRIP1 gene: helps to repair damaged DNA; mutation associated with an increased risk of breast and ovarian cancer.
- CDH1 gene: helps to bind cells in a tissue together; mutation associated with an increased risk of invasive lobular breast cancer.
- CHEK2 gene: provide instruction for protein production and inhibits tumor growth; mutation associated with an increased risk of breast, colon and prostate cancer.

- MRE11A gene: helps to repair damaged DNA; mutation associated with an increased risk of cancer.
- NBN gene: helps to repair damaged DNA; mutation associated with an increased risk of breast and other types of cancer.
- PALB2 gene: involved in the production of protein, DNA repair, and inhibition of tumor growth; mutation associated with an increased risk of breast cancer.
- PTEN gene: regulates cell growth; mutation associated with an increased risk of breast cancer.
- RAD50 gene: helps to repair DNA damage; mutation associated with increased risk of breast cancer.
- RAD51C gene: helps to repair DNA damage; mutation associated with increased risk of breast and ovarian cancer.
- STK11 gene: helps to regulate cell growth; mutation associated with increased risk of breast, lung, and ovarian cancer.
- TP53 gene: involved in the production of protein and inhibition of tumor growth; mutation associated with increased risk of breast cancer and other cancers.

Genetic Testing

There are several tests available to identify individuals who are at risk of developing breast cancer due to the presence of an abnormal gene linked to breast cancer.

These tests are not routinely recommended for the general population but may be offered to individuals who have been diagnosed with breast cancer or have a strong family history of the disease.

There are currently tests available for the following genes: BRCA1, BRCA2, ATM, CDH1, CHEK2, MRE11A, NBN, PALB2, PTEN, RAD50, RAD51C, and TP53.

Reviewed by Susha Cheriyedath, MSc

References

- http://www.breastcancer.org/risk/factors/genetics
- https://ghr.nlm.nih.gov/condition/breast-cancer
- https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fa

ct-sheet
- http://www.cancerresearchuk.org/about-cancer/type/breast-cancer/about/risks/breast-cancer-genes
- https://ww5.komen.org/uploadedFiles/_Komen/Content/About_Breast_Cancer/Tools_and_Resources/Fact_Sheets_and_Breast_Self_Awareness_Cards/Genetics%20and%20Breast%20Cancer.pdf

Further Reading

- What is Breast Cancer?
- Breast Cancer Classification
- Breast Cancer Symptoms
- Breast Cancer Causes
- Breast Cancer Pathophysiology
- Breast Cancer Diagnosis
- Breast Cancer Epidemiology
- Familial Breast Cancer
- Breast Cancer Awareness
- Living with Breast Cancer
- What is Invasive Ductal Breast Cancer?
- Hormone Receptor Positive Breast Cancer
- Preventing Breast Cancer
- Preventing Breast Cancer
- Breast Cancer Management
- Breast Cancer Prognosis
- History of Breast Cancer
- Breast Cancer Society and Culture
- Detecting breast cancer in dense breasts
- What is Breast Reconstruction?
- What is a Mammary Gland?
- What are Aromatase Inhibitors?
- What is the BRCA1 Gene?
- What is the BRCA2 Gene?
- What is the HER2 Gene?
- HER-2 Positive Breast Cancer
- What is Metastatic Breast Cancer?
- What is Ductal Carcinoma in Situ (DCIS)?
- What is Triple Negative Breast Cancer?
- Gail Model (Breast Cancer Risk Assessment Tool)

- Intraductal Papilloma - Benign Tumors of the Breast
- What Does a Quadrantectomy Involve?
- Radial Scar of the Breast
- Radial Scar Diagnosis and Management
- Sentinel Lymph Nodes
- What Does a Sentinel Node Biopsy Involve?
- Treating Breast Oil Cysts
- Axillary Lymph Nodes and Breast Cancer
- Axillary Sampling and Sentinel Node Biopsy Comparison
- Phyllodes Tumors – Follow Up Care

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