strand center
FOR GENOMICS & PERSONALIZED MEDICINE

Your genes know it. Shouldn't you too?

Genetic Tests for Diagnosis Of

Breast Cancer

Your questions answered

Introduction

Breast cancer is one of the most common cancers in women with most cases occurring after the age of 50. It is the second leading cancer amongst women in India, and higher among urban women. The average incidence rate varies from 22-28 per 1,00,000 women per year in urban areas, to 6 per 1,00,000 per year in rural areas. While breast cancer can occur in both men and women, it is far more common in women.

Public support for breast cancer awareness and research funding has helped advance its diagnosis and treatment. Breast cancer survival rates have increased, and the number of deaths has been declining, thanks to a number of factors such as earlier detection, new treatments and a better understanding of the disease. A major cause of concern with breast cancer is that most cancers are detected very late. This results in a high rate of mortality amongst women.

The one thing that every woman should know is that breast cancer is highly treatable especially when detected early. Therefore, it is essential to get familiar with new and important information about breast cancer research and available tests. So, whether you are facing a cancer diagnosis and the challenges of treatment, or wish to prevent the disease if you're at high risk, this booklet answers your questions with reliable and up-to-date information.
It throws light on the genetic basis of breast cancer and the advantages of early screening with advanced genetic diagnostic tests, offering key support for every woman.

Q: What are the different kinds of breast cancer?

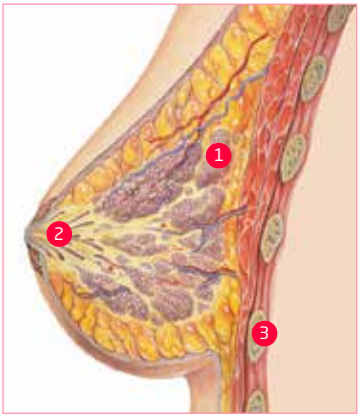
A: Breast tissues are made up of milk glands and milk ducts. Sometimes, some of these cells start dividing abnormally and produce a small group of cells. This group of cells can continue to divide and produce a lump or a small knot of tissue. Breast cancer can be divided into two types:

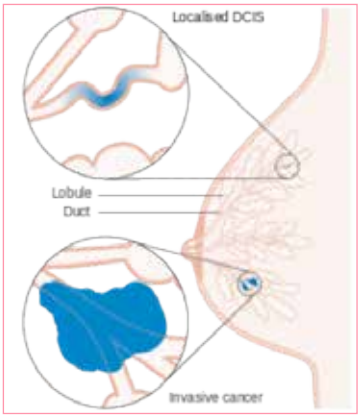
A. Local cancer within the breast

Some people are diagnosed when the cancerous cells are still totally within a duct/lobule. These are known as 'carcinoma in situ'. In Situ refers to the fact that the cancer cells are still located in their place of origin and have not spread to other tissues. Ductal Carcinoma In Situ (DCIS) is the most common type of non-invasive breast cancer.

B. Invasive Breast Cancer

Sometimes, cells can get detached from the original site of growth and can invade neighbouring tissues. Most breast cancers are diagnosed when a tumour has grown from within a duct or lobule into the surrounding breast tissue.
These are called 'invasive' breast cancers. Invasive breast cancers are also divided into those where cancer cells have invaded local blood or lymphatic vessels and those that have not.





1. Milk ducts and lobules
2. Major Ducts
3. Ribs

Q: What is the process of an initial assessment for breast cancer?

A: If you notice a lump or change in your normal breasts, then consult your doctor promptly. The doctor will examine your breasts and armpits to look for any changes. Based on an initial assessment, your doctor may suggest the following:

- Refer you to a specialist
- A biopsy of an obvious lump is sometimes arranged, but other tests may be done first such as:
    - Digital mammogram
    - Ultrasound scan
    - MRI scan
    - Genetic counselling
- Genomic testing and analysis of biopsied tissue if found to be cancerous

Q: What are genomic alterations and what role do they play in cancer?

A: Our cells and tissues work based on instructions provided by our genes. Genes are made up of DNA and are arranged in specific orders on our chromosomes. The DNA that makes up a gene is like a line of code. Usually, the code is fixed and is copied from one cell to another, in an exact manner. Sometimes, there are changes created in this code and essentially mistakes are created in the DNA sequence of a gene. In scientific lingo, these mistakes are known as 'Mutations'. When you have mutations, you have different versions of a gene. This is similar to how we can have different flavours of ice-cream!!

The thing to understand here is that most mutations are harmless. Some mutations can lead to development of serious health issues like cancers. Scientists have examined the associations between mistakes or variations in some genes and certain diseases like breast cancer. We know now, that there are two important genes, namely *BRCA1* and *BRCA2* (pronounced as bra-ca, as a short form of Breast Cancer) that are linked to breast cancer. Changes in the DNA sequence of these genes can cause breast cancer. However, *BRCA1* and *BRCA2* are not the only actors in this drama. At Strand Center, we have identified a set of 13 more genes, and 51 variations in total, in genes other than *BRCA1* and *BRCA2*, in Indian women.

Q: How does this knowledge help women?

A: At Strand Center, we have developed tests to identify mutations that are found to have a strong association with breast cancer. In some cases, we have also looked at genes that are passed from parent-to-child via a family's bloodline. We use this information in two ways:

1. We can use our tests to identify the mutations present in the cancerous tissue. Once the nature of mutation is figured out, this information can help your doctor in choosing the right drugs for the right kind of patient.
2. If we find that many members of a family are falling prey to cancer, our tests can look for specific mutations that can be transmitted from parent-to-child. We then advise members of a family to get tested and confirm whether they do or do not have the mutations and what is their risk of developing breast and other cancers as well.

Q: How can knowing my cancer's genomic information help my treatment?

A: Studies have shown that cancers with some known mutations respond better to certain drugs. If you get tested at the Strand Center, we look for this information in addition to figuring out specific mutations. Taken together, the identity of the mutations and the drugs that can be effective in treating a disease with these mutations, your doctor can chart out a treatment plan for you. In some cases, there are drugs that can attack cells bearing the mutations and spare normal cells and surrounding tissues. These options can be chosen based on knowledge of the type of mutation present in each patient. We have also had cases wherein treatment options were very few and the risk for death was very high. Our tests indicated that genes involved in other diseases were also mutated in these rare cases. Based on such information, doctors were able to choose drugs that helped the patient survive the rare and difficult disease. So, genomic information can help to personalize medicine to some extent and help us in treating cancer in a more specific manner.

Q: Where can I go for help if I'm worried about my risk?

A: If anyone in your family has had breast or ovarian cancer and you're worried about your risk, you can talk to one of our genetic counsellors. Call 1-800-1022-695 (toll free in India) to make an appointment. You can also write to us at stayaheadofcancer@strandls.com.

Q: How do I decide whether to undergo a genetic test for breast cancer or not?

A: We have genetic counsellors who can help you figure out whether your family tree has an increased prevalence of cancer-causing genes. Also, in case you have been diagnosed with cancer, using other diagnostic procedures, you can still talk to us for a genetic analysis. We can figure out the type of cancer that you have and that information can help your doctor make appropriate therapy choices.

Q: I already know I have a family history of breast cancer. Why should I get tested?

A: Testing for inherited risk of cancer helps you and your doctor understand your risk so you can make the best choices for your preventive medical care. Knowing your family history is an important first step, but genetic testing can give you a more accurate picture of your inherited risk.

Q: What if I don't have a family history of breast cancer? Should I be tested anyway?

A: In general, those without a family history do not need to be tested. Individuals who might consider genetic testing include:

- Women with breast cancer before menopause who have had a family member diagnosed with pre-menopausal breast cancer or ovarian cancer at any age
- Women diagnosed with both breast and ovarian cancer
- Someone with a relative who has a *BRCA1* or *BRCA2* mutation
- Men with a personal history of breast cancer

Testing individuals based on the above criteria will also help determine if other family members are at risk for developing breast cancer.

Q: Does a negative test result rule out my risk of breast cancer?

A: A negative test result indicates that you are not at increased risk for breast/ovarian cancer compared to the general population. A negative test also tells you that you are unlikely to suffer from hereditary or early-onset breast cancer. Breast cancer can also occur because of new mutations in breast cells, as a new phenomenon. Therefore, you should continue regular screening measures such as breast self-examination and other check ups.

Q: How do I initiate the process of testing?

A: You can get in touch with our counsellors to understand the process of getting tested.
The process of obtaining a tissue sample is very simple. We require a small sample of blood or saliva to get a test done. In case you have undergone a tissue biopsy, we can also perform the test on a sample of the biopsy.

Q: What is the connection between breast and ovarian cancer?

A: Medical science knows today that the same set of genes are involved in causing breast as well as ovarian cancer. This is particularly true for *BRCA1* and *BRCA2*, two of the better known genes implicated in breast and ovarian cancer. Hence, if our genetic test confirms the presence of *BRCA1* and *BRCA2* mutations, we can estimate the risk of developing either type of cancer in a person.

For more information on how genetic testing can benefit early diagnosis and treatment for breast cancer, visit: www.strandls.com/stayaheadofcancer

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About Strand Life Sciences

A History of Innovative Genomic Research

Strand Life Sciences is a global genomic profiling company aimed at empowering cancer care and genetic testing for inherited diseases. Strand works with oncologists, pathologists, and community hospitals to enable faster clinical decision support for accurate molecular diagnosis, prognosis, therapy recommendations, and clinical trials. Strand's central reference laboratory is located in Bangalore, India.

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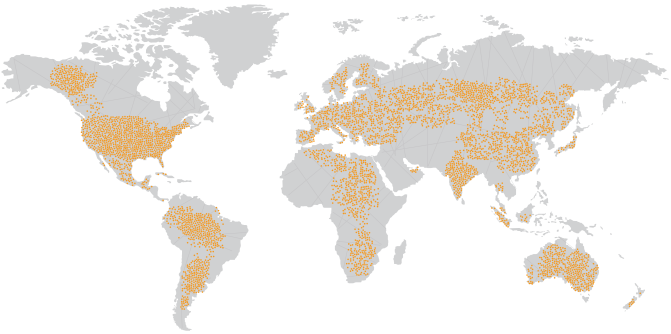

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