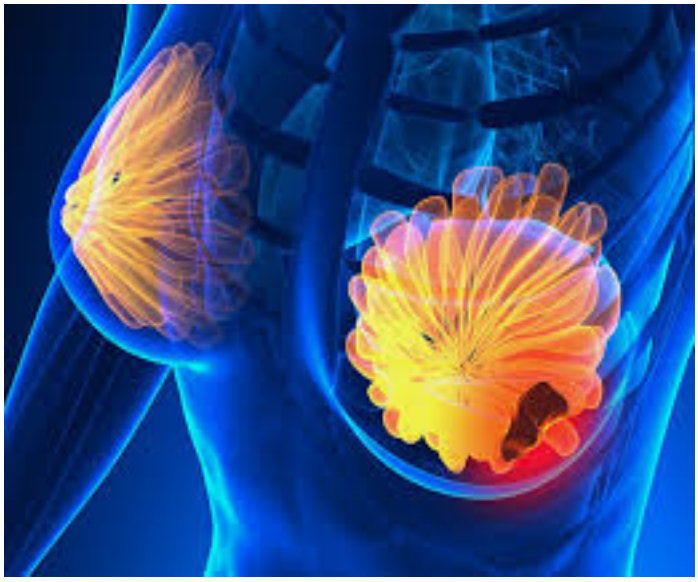
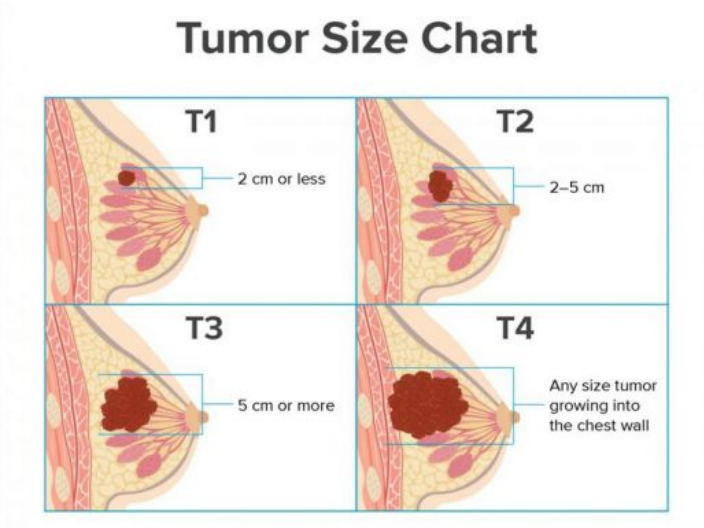
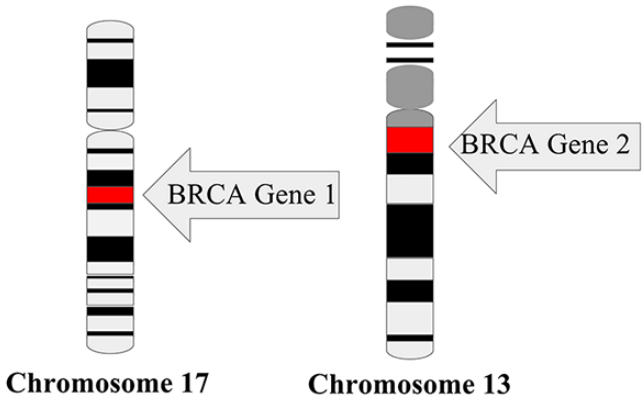
Breast Cancer Genomics







Genomic Testing for Breast Cancer Prognosis & Diagnosis

Breast cancer in women is one of the leading causes of death worldwide and more so in Asia. It can affect men as well. It is estimated that more than 10% of breast cancers are due to genetic inheritance, indicating the necessity for a genetic screening if an individua has a family history of any cancer. The mutant genes of well known Breast Cancer genes *BRCA1* and *BRCA2* can be inherited from either parent, who may be carriers. There are several other genes that are critical for disease prognosis and diagnosis. Recent studies have identified at least 11 novel gene mutations that predispose an individual to Breast Cancer even though he/she is negative for any known BRCA mutations.

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Clinical Genomics

Molecular Genetics

*Mutations in **BRCA1** account for nearly more than 80% of inherited breast and ovarian cancers. BRCA genes form a part of protein complex that stabilizes the genome and controls DNA repair and replication. Hence, mutations in these critical genes fosters more errors in DNA replication and repair leading to more genome-wide mutations leading to several cancers. Recent publications indicate mutations in another gene **Rad51C** may be more critical for Asian women of Chinese ethnicity. **PALB2, BARD1, CHEK2, FANCA, TP53, PTEN, STK11** and **CDH1** gene mutations are some other well known candidate genes for Breast Cancer prognosis. Several other minor variants of lesser significance have been identified that are involved in the disease progression and management. Thus, "cascade genetic screening" for Breast Cancer is advised to individuals with a family history of any cancer*

Pharmacogenomics

- Presence of variant rs4880 in gene ***SOD2*** causes drug toxicity leading to decreased survival when treated with Cyclophosphamide
- Presence of variant rs3892097 in gene ***CYP2D6*** causes decresed survival when treated with Tamoxifen
- Presence of variant rs4870061 in gene ***ESR1*** with TT genotype causes drug toxicity leading to decreased bone density when treated with Letrozole

Why myClinGen?

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