

Hereditary Male Breast Cancer

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1. Introduction

- Breast cancer is the most common malignancy among females and the leading cause of cancer death in relation to women.
- Male breast cancer (MBC) is rare accounting for <1% of all breast cancer cases and its prevalence increases with age.
- About **5-10%** of breast cancer cases are hereditary and are caused by pathogenic mutations in BRCA1 and BRCA2 genes.
- BRCA1 mutations are more common in female breast cancer whereas male breast cancer is characterized by BRCA2 mutations although women can have BRCA2 mutations and men BRCA1 mutations.
- There are some different types of treatment for men with breast cancer like surgery, chemotherapy, radiotherapy and more others.

2. Material and Methods

- Literature research using mainly three free databases: Medline Pubmed, Uptodate and Elsevier. The parameters or the key words sought were:
 - Breast cancer
 - Male
 - Hereditary

3. Male Breast Cancer

- Male breast cancer (MBC) is rare in contrast to female breast cancer and this has been reflected in research. It has an Incidence rate of **0.5/100000** men per year, **0.1%** of male cancer deaths.
- Usually appears in men 60 to 70 years.
- Risk factors:
 - Gynecomastia
 - Testicular conditions
 - Klinefelter syndrome
- Inherited mutations in BRCA also increase the risk of MBC and it appears to be higher with inherited BRCA2 rather than BRCA1 mutations.
- There are 5 stages of MBC.

4. Genes and signalling pathways involved

- Approximately, a **45%** of families with hereditary breast cancer have mutations in BRCA genes.
- Hereditary breast cancer occurs in families in which there are some family members affected of different generations.
- Only **5%** of all hereditary breast cancer are due to BRCA1 and BRCA2 mutations.
- Male BRCA2 mutations carriers confer a lifetime risk of breast cancer (6%).
- BRCA2 related tumors usually express estrogen and progesterone receptors.

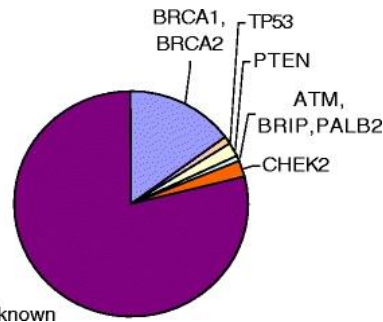


Fig 1. Genes involved in hereditary breast cancer. *Cell Oncol (Dordr)* 2011 April; 34(2): 71–88. Published online 2011 February 19. doi: 10.1007/s13402-011-0010-3

5. Diagnosis

Clinical diagnosis	- Mammography - Biopsy
Differential diagnosis	- Includes gynecomastia, breast abscess, metastases to the breast and other non-breast cancer primary tumors.
Molecular diagnosis of BRCA1 and BRCA2 genes	- Targeted mutation analysis - Sequence analysis - Deletion/duplication analysis

Table 1. Diagnosis for MBC.

5. Differences between Male and Female

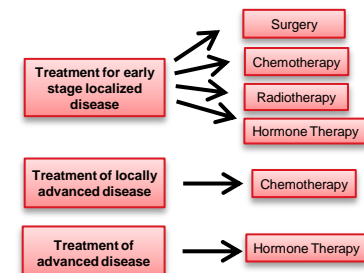
Characteristics	MBC	FBC
Incidence rate	0.5-1/100000 men per year	1-4 million of annual new cases
Main gene mutation	BRCA2	BRCA1
Estrogen receptor or progesterone receptor	or 90% of MBC	60-70% of FBC
Prognosis	Worse due to older age and more advanced disease stage at diagnosis	Better due to early age of diagnosis
Treatment	Mastectomy as main treatment	Mastectomy as main treatment

Table 2. Differences between Male and Female.

* MBC (Male Breast Cancer), FBC (Female Breast Cancer).

6. Treatment

- Some of the treatments are standards and very similar to those used in women while others are being tested in clinical trials. Patient participation in clinical trials are often the best way to treat cancer and should take into account when choosing the treatment.
- Breast cancer treatment is divided in 3 groups according to the disease stage:



CONCLUSIONS

- ✓ Male breast cancer is rare compared to female
- ✓ Only 5-10% of breast cancer are hereditary
- ✓ Risk factors like gynecomastia can cause male breast cancer.

- ✓ BRCA2 affects 40% of men with breast cancer
- ✓ The diagnosis is essential when there is suspicion of breast cancer.
- ✓ Treatment is similar in male and female.

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