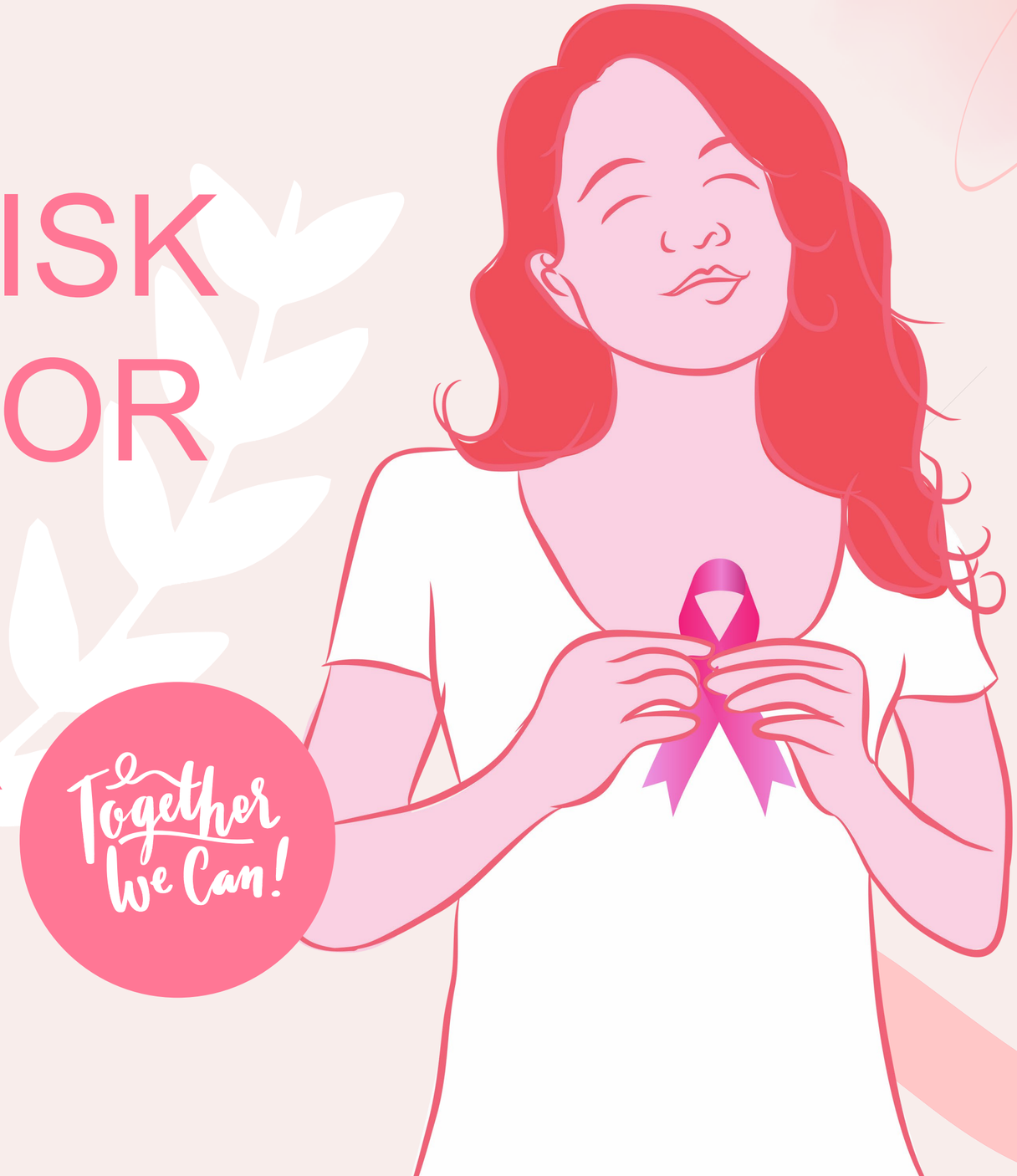
GENETIC RISK FACTORS FOR BREAST CANCER

Awareness

Pr es entation



Group:
C13

Objectives

- Trace the historical development of genetic risk factor identification and testing in breast cancer
- Differentiate between the major breast cancer related genes and their associated risk profiles
- Analyze current trends and predict potential future advancements in genetic risk factor assessment for breast cancer
- Evaluate the potential psychological and ethical implications of patients’ awareness of their genetic breast cancer risk factors

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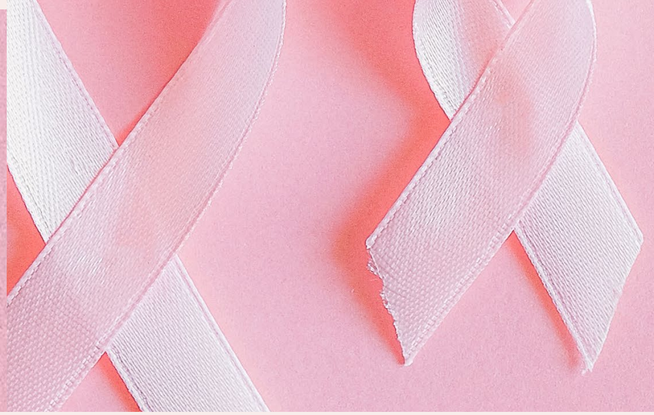
History of Genetic Risk Factors



History of Genetic Risk Factors

1970's : American geneticist Mary -Claire King began researching the cause of breast cancer that runs in families. She hypothesized that a genetic mutation was responsible for some forms of familial breast cancer.

1990's : First genetic risk factors for breast cancer were discovered with the BRCA1 and BRCA 2 gene.

History of Genetic Testing for Breast Cancer

History of Genetic Testing for Breast Cancer

1996

BRCA1/2 mutation screening became available as a clinical service.

1997

The University of Pennsylvania became the first organization in the United States to offer the BRCA 1/2 test for clinical purposes.

1997

UCSF-CGPP receives IRB approval for long -term follow -up program - to promote research efforts associated with hereditary cancer risk.

June 2013

U.S. Supreme Court invalidates specific claims made by Myriad Genetics.

June 2013

Allowed other companies to begin offering panel tests that included the BRCA 1/2 genes.

2018

23andMe received FDA approval to identify three variants in the BRCA 1/2 genes.

BRCA1 & BRCA2 Gene

BRCA 1 & BRCA 2

- Function: tumor suppressor that help regulate cell division.
- Inheritance: people inherit one copy of each gene from each parent.
- Risk: People with the mutations in these genes have a higher risk of developing breast and ovarian cancers.
- Age of Onset: People with mutations in these genes tend to develop at younger ages.
- Treatment: Increased surveillance, chemoprevention, and prophylactic mastectomy or oophorectomy.

ATM Gene

Ataxia - Telangectasia Mutated (ATM) Gene

- Ataxia -Telangiectasia are frequency detected in Breast Cancer with an incidence ranging up to 40%.
- Ataxia -Telangectasia Mutated (ATM): some variant are associated with an increased risk of Breast Cancer and worse prognosis.
- ATM gene is considered a “cancer protection” gene because it helps agains breast, prostate, and pancreatic.
- Bladder, Breast, Melanoma, Stomach, Pancreas, Lung, and Ovary cancer is all associated with ATM.

Other Genes

Other Genes

BARD1, BRIP1, CASP8, CYP19A1, FGFR2, H19, LSP1, MAP3K1, MRE11A, NBN, RAD51, and TERT.

There is also a gene mutation called Cowden Syndrome that links to breast cancer, uterine, and thyroid cancer.

What Do All These Gene’s Mean?

Overall,

While we can’t change our genetics or family history of cancer, knowing that you are at a higher risk can help with creating an early detection plan to detect breast cancer in it’s earliest stages, while it is still localized .

How Does This Effect a Mammogram?

Generally...

- Having any positive genetic risk factors doesn’t change anything for your mammogram. Typically, you may just start screenings a little earlier in your life.

- Just because you are postive for any of the genetic genes, doesn’t mean you are gauranteed to have breast cancer!

According to National Health Service of The United Kingdoms....

Out of every 100 women who have a BRCA 1 gene mutation:

- 65 - 85 will develop breast cancer in their lifetime
- 40 - 63 will develop ovarian cancer in their lifetime

The Future

The Future...

More personalized treatment plans. Genetic testing of tumor tissue has the potential to identify different variants, thus be able to identify more people who might benefit from target therapies.

Benefit’s For The Patient (of Knowing)

Piece of Mind

Family History

Treatment Options

Cancer Risk Management

Early Detection

Harmfulness To The Patient (of Knowing)

Psychological Stress

“Survivor’s Guilt”

Uncertainty

Cost

Privacy and Discrimination Issues

Possibility of Rare False-Positive

Conclusion

Overall...

The advancements in genetic testing and genetic risk factors for Breast Cancer can help a patient if they want to learn more about it. Some patient’s want to know and some do not and respecting either choice is very important for the mammographer.

Thank you!

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