PREVALENCE OF PATHOGENIC/LIKELY PATHOGENIC (P/LP) MUTATIONS AMONG BREAST CANCER PATIENTS PRESENTING TO A TERTIARY CARE CENTRE IN SOUTH INDIA.

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BACKGROUND OF THE STUDY:

- Genetic testing for breast cancer has become an essential tool in identifying individuals at increased risk, guiding treatment decisions, and informing preventive measures.
- BRCA1 and BRCA2. Mutations can increase the risk of developing breast cancer and Identifying individuals with pathogenic mutations allows for personalized risk assessment and surveillance.

OBJECTIVES:

- Study the prevalence of P/LP mutations in breast cancer patients.
- Describe the profile of P/LP mutations in high-risk individuals.

METHODOLOGY:

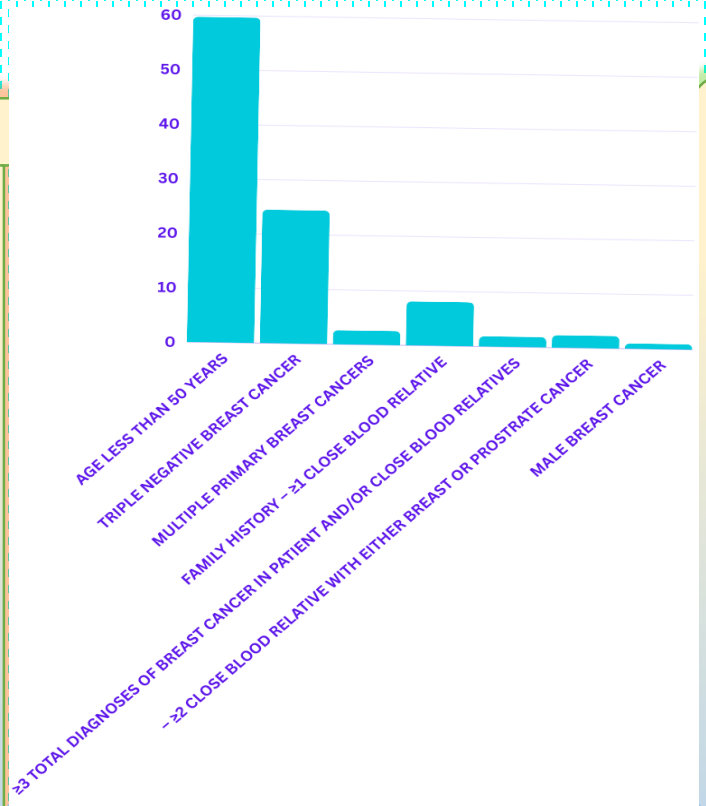
- All breast cancer patients eligible for genetic testing were counselled and offered genetic testing.
- Targeted multigene sequencing was done using Next generation sequencing in which143 mutations were tested.
- Post-test counseling was done for patients and family members including cascade testing.

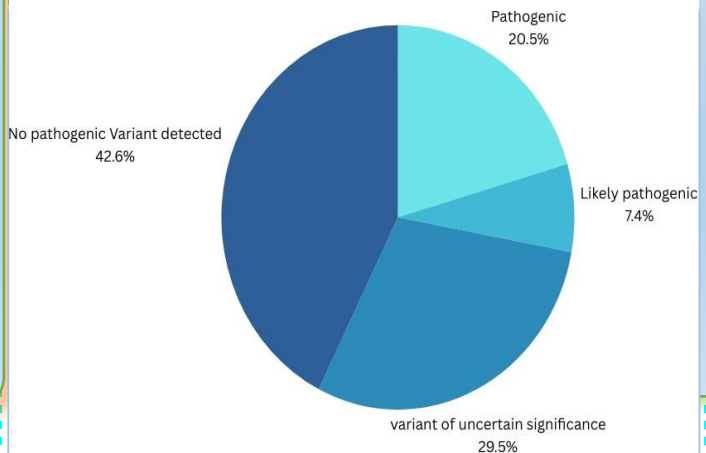
RESULTS:

Data from November 2022 to February 2024

Number Of Patients Counseled – 428.

- 409 breast cancer patients
- 19 high risk relatives
- 248 consented for testing
- 190 test reports available; 58 awaited
- 5 patients had 3 risk factors
- 55 patients had at least two risk factors





Prevalence Of P/Lp Mutations:

- 25 - BRCA1 11 - BRCA2
- High penetrance genes (BRCA1, BRCA2, TP53, PTEN, CDH1, STK11)- 37 (71%)
- Moderate penetrance genes (PALB2, BRIP1, CHEK2, ATM, BARD1)- 9 (17%)
- Other genes MLH1, NF1, RAD51D, CHEKC, MUTYH, BLM, FANCM, NBN

Number Of Indications And Prevalence:

- Overall prevalence of P/LP mutations- 27.9%
- Prevalence in patients with 2 indications of genetic testing- 47% (26/55)
- 3 indications- 80% (BRCA1)

Cascade Testing:

- 154 at risk relatives were identified. Only 20 relatives turned up for counselling.
- 9 opted for testing
- 3 showed BRCA1 mutations.
- Others had no variants

CONCLUSIONS AND RECOMMENDATIONS:

- P/LP mutations are common (28%) among high risk breast cancer patients
- Multi gene panel testing is recommended, as a significant proportion (31%) of patients have non BRCA mutations.
- Novel strategies like physician assisted cascade testing by other health care professionals is recommended to increase cascade testing.