PREVALENCE OF BRCA1 AND BRCA2 MUTATIONS WITH EARLY-ONSET BREAST CANCER

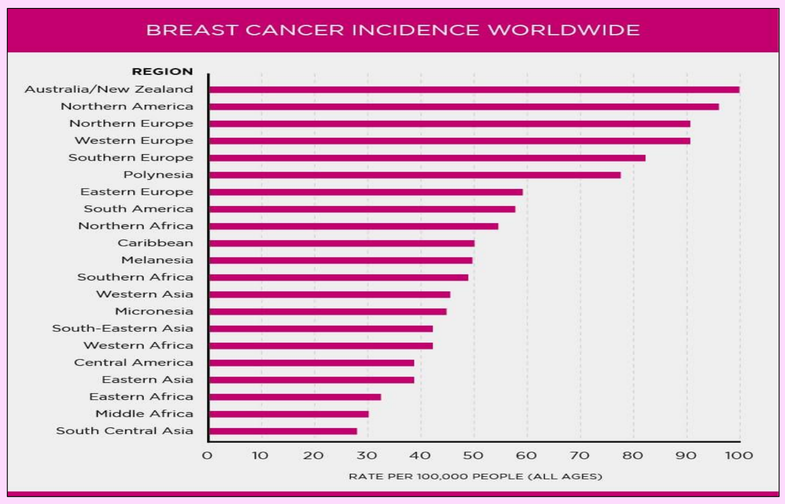
Roha Tariq[1], Irfan Ishaque[2], Ariba Shah[3]
1. Department of Biotechnology, Lahore College for Women University, Pakistan
2. Department of Zoology, Government College University, Lahore, Pakistan
3. Ziauddin University, Karachi, Pakistan

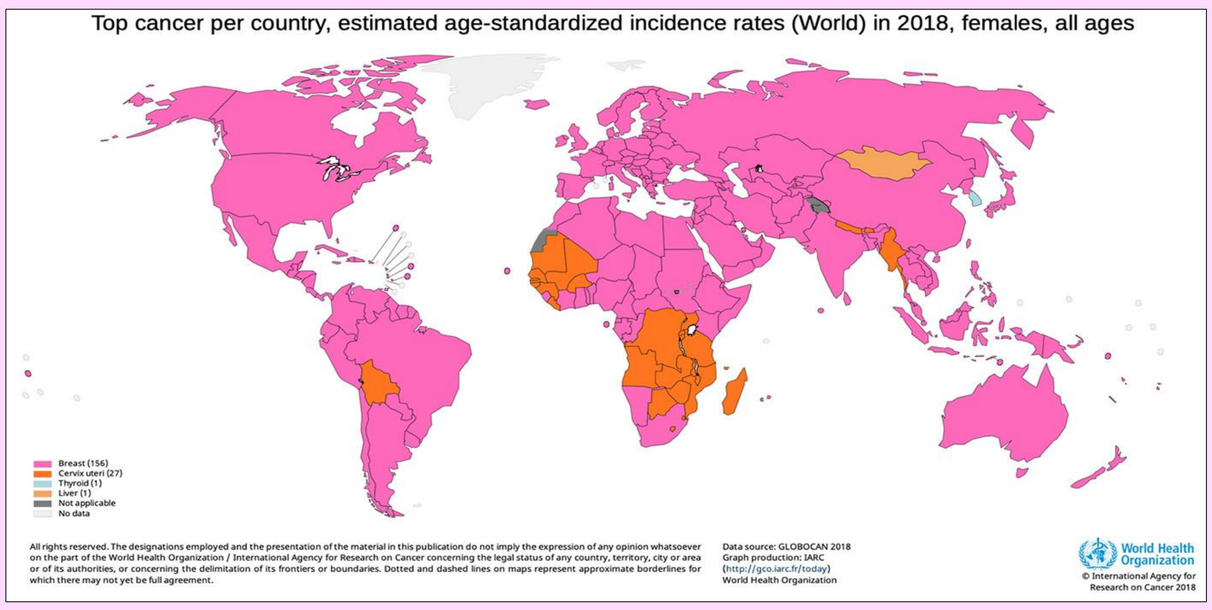
Introduction:

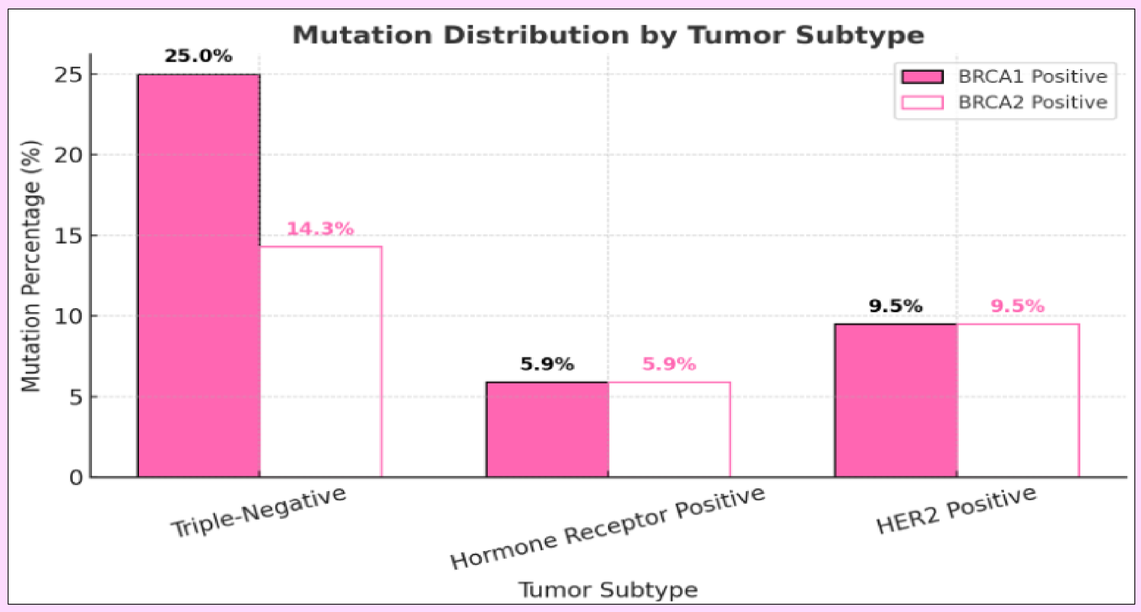
Early-onset breast cancer poses significant clinical challenges due to its aggressive behavior and genetic predisposition. Mutations in the BRCA1 and BRCA2 genes are strongly linked to hereditary breast cancer, especially in younger patients. However, regional data on their prevalence remains limited in South Asian populations, particularly in Pakistan. The major objective of this study is to determine the prevalence of BRCA1 and BRCA2 gene mutations among women diagnosed with early-onset breast cancer in the Lahore region of Pakistan.

Methods:

A cross-sectional study was conducted over eight months involving 200 women aged ≤39 years with histologically confirmed breast cancer. Participants were recruited from three major oncology centers in Lahore. Clinical and demographic data were collected through structured interviews. Peripheral blood samples were analyzed using next-generation sequencing (NGS) for BRCA 1/2 mutations. Statistical analysis was performed by SPSS v26, with significance set at $p<0.05$.

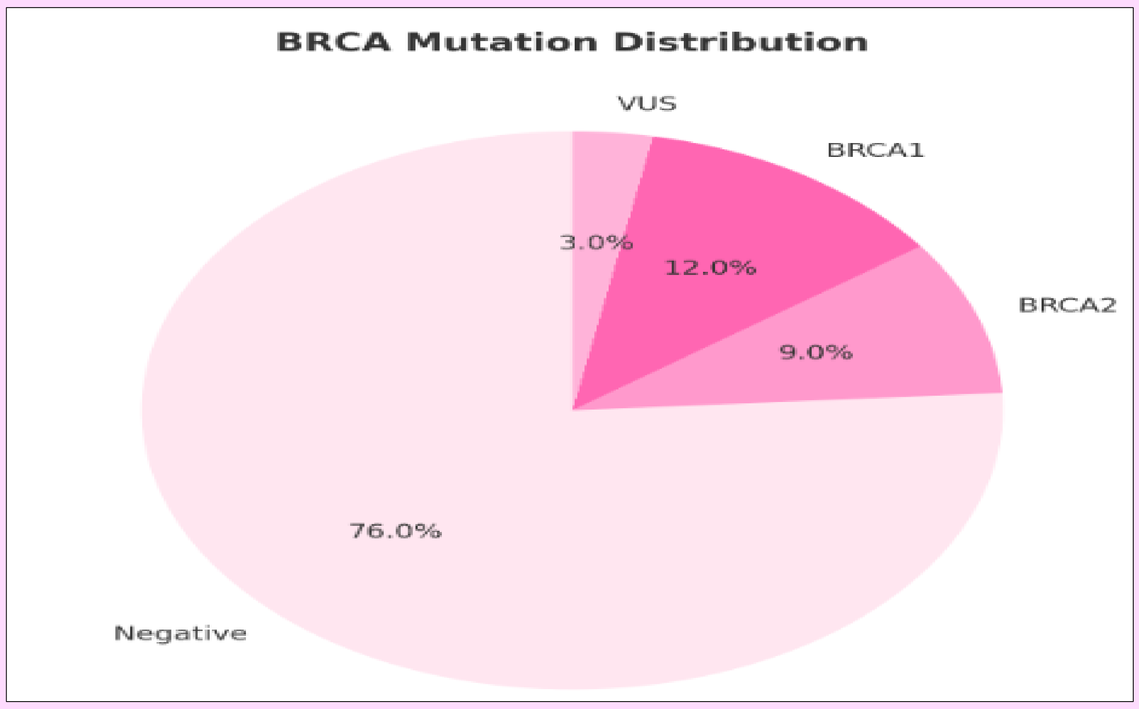


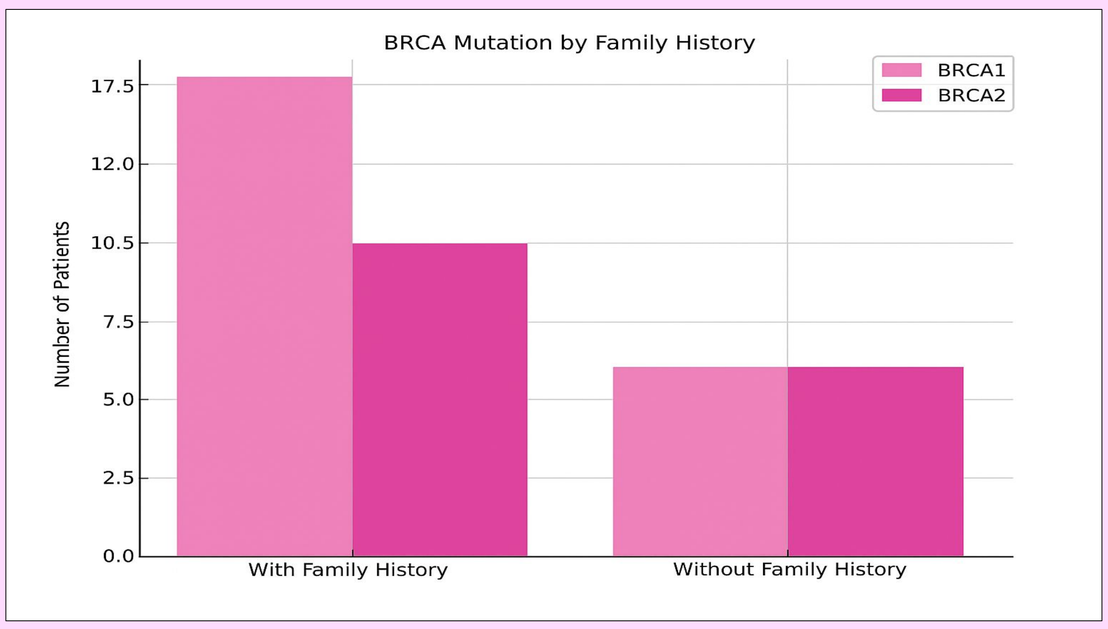




Results:

BRCA1 mutations were identified in 24 patients (12%) and BRCA2 mutations in 18 (9%). Variants of uncertain significance were found in 6 patients (3%), while 152 (76%) tested negative for both mutations. Mutation prevalence was significantly higher in patients with a family history of breast or ovarian cancer and among those with triple-negative breast cancer. BRCA1 mutations were most common in triple-negative cases (25%), while BRCA2 mutations showed a slightly more even distribution across subtypes.





Conclusion:

A considerable proportion of early-onset breast cancer patients in Pakistan carry BRCA mutations. These findings underscore the importance of routine genetic screening and tailored risk management strategies in young breast cancer patients, irrespective of family history.

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