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Awareness and Determinants of Uptake of Genetic Testing Among Breast Cancer Patients and their Family Members: An Exploratory Mixed-Methods Study

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Abstract

Background

Genetic testing is becoming increasingly essential in breast cancer because it can inform treatment decisions, detect hereditary cancer risk, and assist family members who may benefit from early risk assessment and cascade testing. However, the adoption of genetic testing is dependent on patients' awareness, comprehension, desire, and access to counseling and testing services, all of which may be hampered by practical constraints in India. This study looked at the awareness, beliefs, and variables that influence genetic testing among breast cancer patients and their first- and second-degree relatives.

Methods

An exploratory mixed-methods study was carried out at the National Cancer Institute, AIIMS, New Delhi. Twenty participants were interviewed, out of which 10 were breast cancer patients and 10 were first- or second-degree relatives. A questionnaire was administered to another 20 participants, 10 patients and 10 family members, respectively, including questions on socio-demographics, awareness and willingness to genetic testing, barriers, etc. Thematic analysis was used for qualitative data, and descriptive

statistics were used to analyze quantitative data. Results from both components were combined.

Results

The qualitative findings indicated significant gaps in knowledge and comprehension of genetic testing, including misunderstanding between genetic testing and diagnostic procedures like biopsies. Despite their limited understanding, participants had positive opinions regarding testing, which was mostly due to confidence in healthcare personnel and family support. Cost, distance, restricted testing availability, and insufficient provider communication were among the major hurdles. Quantitative findings supported high desire and communication gaps: 90% of patients were willing or potentially willing to undergo testing if advised by a doctor, and 100% of attendants would advocate testing. 70% of patients described cost as a key obstacle, 90% indicated worry or concern about their genetic risk, and just 40% had addressed genetic risk with a healthcare professional.

Conclusion

Despite the lack of awareness, breast cancer patients and their families expressed a high willingness to undergo genetic testing. Improving provider communication, genetic counseling, and

availability to low-cost testing may help turn willingness into informed action.

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