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Perspectives on PGT-M in Hereditary Breast Cancer: Insights from Japanese Patients Undergoing Fertility Preservation

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Abstract

Introduction:

Preimplantation genetic testing for monogenic disorders (PGT-M) offers BRCA variant carriers an option to prevent transmission of hereditary cancer. While its global application is expanding, PGT-M has not yet been formally approved or implemented for hereditary cancer syndromes in Japan.

Objectives：

This study investigates awareness and attitudes toward PGT-M among breast cancer patients who underwent fertility preservation.

Methodology:

A questionnaire-based survey was conducted among 264 breast cancer patients eligible for oocyte or embryo cryopreservation at IVF clinics between October 2024 and March 2025. A total of 159 valid responses were analyzed. The survey assessed BRCA testing status, PGT-M awareness, willingness to undergo PGT-M, and opinions on future availability.

Results:

BRCA1/2 testing uptake was 53.5%; 18% of respondents were variant carriers. Only 16.4% had prior awareness of PGT-M, and 48.4% expressed willingness to use PGT-M if available. Among BRCA-positive patients, 36.4% considered using PGT-M, and 72.7% believed it should be made available upon request. Overall, 69% supported information sharing between oncology and fertility providers.

Conclusion:

This is the first patient-centered survey in Japan on PGT-M for hereditary cancer syndromes. The findings highlight the importance of expanding both reproductive options

and patient awareness of PGT-M as part of survivorship care in hereditary cancer patients. Moving forward, discussions should focus on how best to provide accurate information and enable informed reproductive choices for those at genetic risk.