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妊孕性温存療法を実施した乳癌患者を対象とした着床前遺伝学的検査に対する意識調査

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

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Background:
Preimplantation genetic testing for monogenic disorders (PGT-M) offers BRCA variant carriers an option to prevent the 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. This study investigates awareness and attitudes toward PGT-M among breast cancer patients who underwent fertility preservation.

Methods:
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.