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Current status of BRCA mutations in breast cancer patients treated with fertility preservation therapy at our clinic

BRCA1/2 mutations trigger Hereditary breast and ovarian cancer syndrome (HBOC). In Japan, genetic testing for BRCA1/2 was covered by insurance in 2018, and the number of tests is increasing. Fertility preservation therapy before cancer treatment is an established treatment. Preimplantation genetic testing for monogenic diseases (PGT-M) for HBOC is not yet approved in Japan. In this report, we present the current status of cases in which fertility preservation treatment was performed on female breast cancer patients between January 2018 and April 2024 at our hospital, based on a retrospective review of medical records.

Fertility preservation therapy was performed on 88 patients during the period, with an average age of 35.9 years. 76 actually underwent treatment. After cancer treatment, 15 patients underwent embryo transfer and 9 patient had live births. The median period from freezing to transfer was 41 months. The number of patients to determine whether or not BRCA1/2 genetic testing had been

performed was only 13 (2 positive, 5 negative, 3 with unknown results, and 3 without testing) of the 76 women who underwent fertility preservation. 12 out of 13 patients started cancer treatment in 2020 or later.

As the BRCA1/2 tests are increasing, the discussion of PGT-M for HBOC will advance. In the future, based on the guidelines for genetic testing and diagnosis, it will be necessary to make preparations, including surveys of patient awareness and actual conditions, in cooperation with cancer treatment facilities.