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Status of genetic test for hereditary breast and ovarian cancer in Tokyo Women's Medical University, Adachi Medical Center

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Introduction

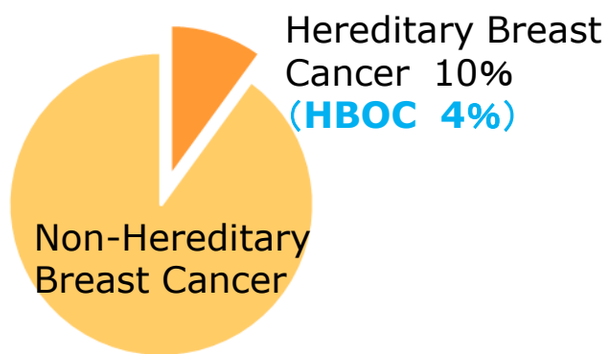
- Approximately 10%[1)] of all breast cancers are hereditary breast cancers, and approximately 4% are hereditary breast and ovarian cancers (HBOC)[2)] caused by the *BRCA1/2* gene.
- In Japan, health insurance made it possible to have *BRCA1/2* genetic test as companion diagnosis for PARP (poly ADP-ribose polymerase) inhibitors in 2018. Diagnosis of HBOC, surveillance and contralateral risk-reducing mastectomy were covered by health insurance. The number of patients undergoing genetic tests increased steadily in 2020.
- This study aimed to summarize the status of testing at our hospital.

Table 1. Insurance coverage conditions for a genetic test for HBOC in Japan

Breast cancer diagnosed at or before age 45 years
Triple-negative breast cancer diagnosed at or before age 60 years
Multiple primary breast cancers in either one or both breasts
Ovarian, fallopian tube, peritoneal cancer, pancreatic cancer and/or male breast cancer
A family member with a known BRCA1/2 pathogenic variant
Three or more relatives with breast cancer at any age

JOHBOC, hereditary breast and ovarian cancers (HBOC) Guidelines 2024

Figure 1.
Patients with Hereditary breast cancer



Materials and methods

- From June 2018 to November 2023, 93 breast cancer patients with *BRCA1/2* genetic test participated in this study.
- The following data were collected from medical records and examined retrospectively.
  Number of genetic tests / Number of items covered by insurance / Timing of genetic tests

Results

Table 2. Characteristics of breast cancer patients who underwent genetic test

Characteristic	N (%)
Total	93
Female	91 (97.8%)
Pathological variant	12 (12.9%)
BRACAnalysis for HBOC	71 (76.3%)
BRACAnalysis for Companion diagnosis	22 (23.7%)

	HBOC	Companion	Total
BRCA1 (+)	4/71 (5.6%)	1/22 (4.5%)	5/93 (5.4%)
BRCA2 (+)	6/71 (8.5%)	1/22 (4.5%)	7/93 (7.5%)

	Luminal type	Triple negative type
BRCA1 (+)	2/7	5/7
BRCA2 (+)	3/5	2/5

Figure 2. Number of genetic tests

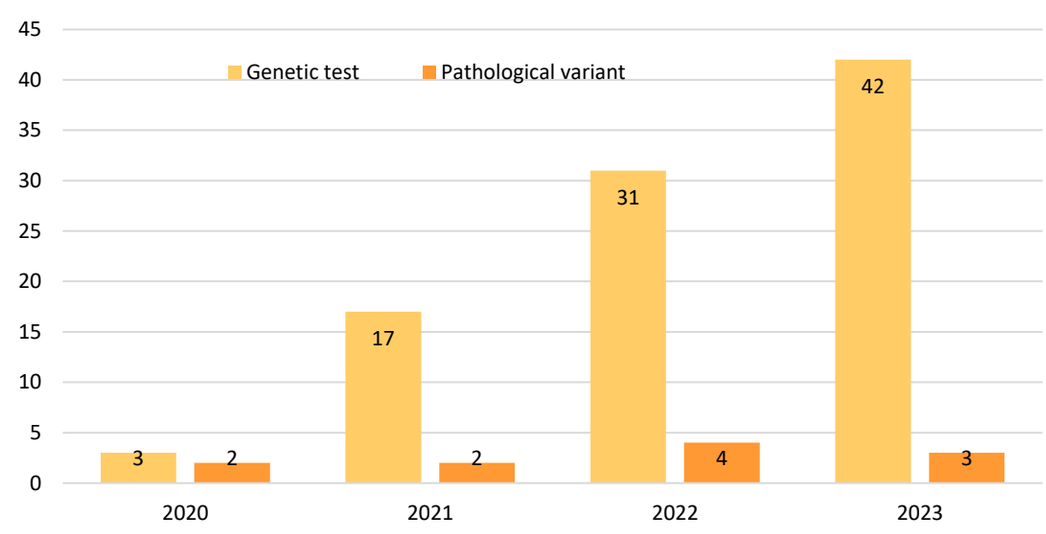


Figure 3.
Number of items covered by insurance

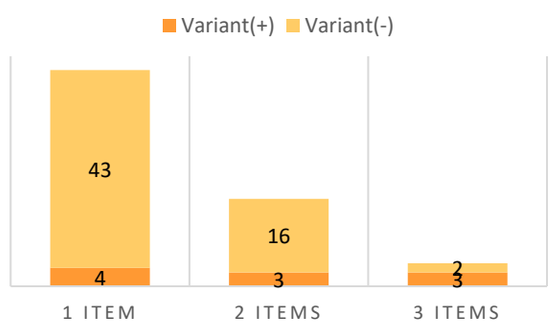
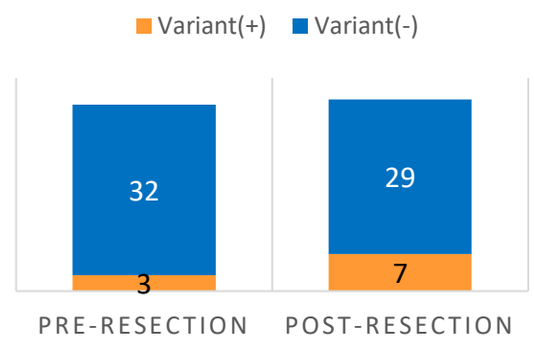


Figure 4.
Timing of genetic tests



Discussion / Conclusion

- The number of *BRCA1/2* genetic tests is increasing, with appropriately selected clients needing to be picked up in our hospital.
- Pre-resection test for HBOC diagnosis allows for surgical decision-making.
- Not only breast surgeons but also clinical geneticists and certified genetic counsellors provide explanations of HBOC.
- We will provide information on hereditary breast cancer by conducting putting surveillance and risk-reducing mastectomy systems.

References
1)*BRCA1*- and *BRCA2*-Associated Hereditary Breast and Ovarian Cancer. In: GeneReviews at GeneTests: Medical Genetics Information Resource.
2)Monozawa Y et al. Germline pathogenic variants of 11 breast cancer genes in 7,051 Japanese patients and 11,241 controls. Nature Communications, 2018; 9(1): 4083

We have no conflicts of interest to declare on this presentation.