

High rate of occult cancer found in prophylactic mastectomy specimens despite thorough presurgical assessment with MRI and ultrasound: findings from the Hereditary Breast and Ovarian Cancer Registration 2016 in Japan.

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Abstract

PURPOSE: Prophylactic surgery is a preemptive strategy for hereditary breast and ovarian cancer (HBOC). Prophylactic mastectomy (PM) reduces breast cancer risk by > 90%. The aim of our study is to analyze the information of the Japanese pedigrees and to utilize the results for clinical practice.

METHODS: We statistically analyzed records of HBOC registrees who had undergone BRCA1/2 genetic testing at seven medical institutions up until 2016. In the cases of PM, we examined breasts with the use of mammography (MMG), ultrasound (US), and magnetic resonance imaging (MRI) before surgery. After PM, the specimens were divided about 1 cm serially and examined in their entirety.

RESULTS: Of 1527 registrees who underwent BRCA testing, 1125 (73.7%) were negative for BRCA1/2 mutation, 297 (19.5%) were positive for BRCA1/2 mutation (BRCA1/2^{MUT+}), and 105 (6.9%) had uncertain results. To decide whether to undergo total mastectomy vs. breast-conserving surgery (BCS), 370 registrees underwent presurgical genetic testing. During the follow-up period, four new-onset breast cancers were found among the 55 non-affected BRCA carriers. Among the 73 BRCA1/2^{MUT+} carriers who underwent BCS, 3 were found to have ipsilateral breast cancer. Of 189 BRCA1/2^{MUT+} carriers with unilateral breast cancer, 8 were found to have contralateral breast cancer. Of 53 PM specimens, 6 (11.3%) were found to have occult breast cancer despite using MMG, US, and MRI.

CONCLUSIONS: Our report showed a relatively higher incidence rate of occult cancer at 11.3% in PM specimens despite thorough pre-operative radiological evaluations, which included a breast MRI. Considering the occult cancer rates and the various pathological methods of our study and published studies, we propose the necessity of a histopathological protocol.

KEYWORDS: BRCA; Hereditary breast and ovarian cancer syndrome; Magnetic resonance imaging; Occult cancer; Pathological method; Prophylactic mastectomy