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From Genetic Testing to Treatment and Prevention of BRCA-Related Breast Cancer

More than one-third of all Koreans are estimated to have cancer from birth to life expectancy, and the cumulative risk of cancer development during their lifetime is 38.3% for men and 33.3% for women. In 2019, 221,347 Korean individuals are expected to be newly diagnosed as having cancer and approximately 82,344 are expected to die due to cancer. Breast and lung cancers are the most common types in women (23.8%) and men (16.4%), respectively [1].

The incidence of breast cancer in Korean women is the highest among women in their fifth decade, while in American women, the incidence is highest among those in their mid-sixth decade or later [2]. One explanation for this difference may be genetic factors. Indeed, several breast cancer-related genes, including *ATM*, *BRCA1*, *BRCA2*, *CDH1*, *CHEK2*, *PALB2*, *STK11*, and *TP53*, have been identified [3]. Among these, *BRCA1/2* are the most important and most widely studied genetic risk factors for breast cancer worldwide.

In this issue of Ann Lab Med, Yoo, *et al.* [4] report the results of genetic testing of *BRCA1/2* and the clinical validity of next-generation sequencing-based multi-gene panel testing in a single institution. Although pathogenic variants (PV) were identified not only in *BRCA1/2* but also in *MSH2*, *PMS2*, *CHEK2*, and *PALB2*, the proportion of PV in *BRCA1/2* was estimated to be over 88%, indicating the need to focus genetic testing and management efforts more on these two genes.

Recently, the US Preventive Services Task Force updated its recommendations on risk assessment, genetic counseling, and genetic testing for BRCA-related cancers in women as follows:

“primary care clinicians assess women with a personal or family history of breast, ovarian, tubal, or peritoneal cancer or who have an ancestry associated with *BRCA1/2* gene mutations with an appropriate brief familial risk assessment tool. Women with a positive result on the risk assessment tool should receive genetic counseling and, if indicated after counseling, genetic testing. (B recommendation)” [5].

This issue of Ann Lab Med also presents a review article by Lee, *et al.* [6], which outlines treatment and prevention strategies for patients with BRCA-related breast cancer. It also covers recent advances in the management of BRCA-related breast cancer by treatment with poly (ADP-ribose) polymerase (PARP) inhibitors.

Conflicts of Interest

None declared.

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