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*Abstract*

# Characteristics of breast cancer patients tested for germline BRCA1/2 mutation at Ramathibodi Hospital during 2014-2018

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**Background:** Germline mutations in breast cancer susceptibility genes (BRCA1 and BRCA2) are the most common causes of hereditary breast and ovarian cancer (HBOC) syndrome. The objective of this study is to assess the characteristics of breast cancer patients with germline BRCA1/2 mutation in our institute.

**Methods:** We retrospectively reviewed 67 breast cancer patients suspected of HBOC who had been tested for germline BRCA1/2 mutation at our Center for Medical Genomics during 2014-2018. BRCA mutations were tested by next-generation sequencing and confirmed by Sanger sequencing. To compare the characteristics of BRCA carriers and non-carriers, we used Fisher's exact test and one-way Analysis of Variance (ANOVA). To identify prognostic factors of disease-free survival (DFS), a Cox regression analysis was performed.

**Results:** A total of 67 patients were analyzed. The results were as follows: BRCA carriers 17.92% (n=12), non-carriers 76.12% (n=51), Variant of Uncertain Significance (VUS) 5.97% (n=4). The mean age at diagnosis was 41.97±10.29 years, 39.83±7.36 years in BRCA1, and 46.17±16.04 in BRCA2. All tumors of BRCA carriers were only the luminal-B subtype, with the luminal-B subtype HER2 positive in two tumors. Unexpectedly, the triple-negative breast cancer (TNBC) subtype was not detected in the tumors of our BRCA carriers. BRCA carriers had worse 3-year DFS, compared with non-carriers (81.48% vs. 90.27%, HR 2.04 (0.64, 6.49), p=0.229). The 3-year DFS was 100%, 84.77%, and 80.00% for stage I, II, and III, respectively. Early-onset patients (□ 40 years) had non-significant higher 3-year DFS (90.78% vs.79.44%, p=0.741), compared with the late-onset patients.

**Conclusions:** This study showed that breast cancers in our BRCA carriers were only the luminal-B subtype and the BRCA carriers had a trend of worse prognosis than non-carriers.