Background: The purpose of this study is to estimate 5-year survival rates for patients with early onset breast cancer, with and without a BRCA1/2, CHEK2, NOD2 mutation or TP53 polymorphisms and to identify prognostic factors among mutation carriers in breast cancer patients.

Methods: In a study conducted in the years 2007–2016 in the Maria Skłodowska Curie Memorial Cancer Centre and Institute of Oncology, Gliwice Branch (CO1) were analyzed prognostic factors and survival in 622 breast cancer including 60 BRCA mutation carriers, 46 CHEK2 mutation carriers, 29 NOD2 mutation carriers and 87 patients with TP53 polymorphisms. Control group was selected from breast cancer patients without mutation and polymorphisms (n = 400).

Results: The five–year rate of OS was 75.9% for pts with BRCA mutation, 94.4% for CHEK2 mutation carriers, 96.6% for NOD2 mutation carriers and 100% for patients with TP53 polymorphisms. BRCA mutation carriers had insignificantly worse survival as compared to control group (p = 0.180). Patients with CHEK2 mutation had significantly better OS than control group (p = 0.032). Similarly NOD2 mutation carriers had also significantly better OS than control group (p = 0.043). Patients with TP53 polymorphisms carriers had higher OS in comparison to control group (p = 0.002). In subgroup of pts with N0 (Without lymph node metastases) BRCA mutation carriers was characterized by the worst OS (81.1%) among carriers of other mutations: CHEK2 (94.7%, p = 0.021), NOD2 (95.3%, p = 0.092) and TP53 polymorphisms (100%, p = 0.007) or control group (94.4%, p = 0.022). Similar tendency was observed according to N+ subgroup and subgroup with tumor size T1-2. Higher tumor size (HR = 2.85), lymph node metastases (HR = 2.93) and HER2 overexpression (HR = 1.49) were significant factors for worse OS. Positive ER status was associated with a better OS (HR = 0.52, p = 0.001). Age <40 years (HR = 0.71, p = 0.253) was insignificantly favorable factor.

Conclusions: CHEK2, NOD2 mutation carriers and patients with TP53 polymorphisms had better 5-year survival in comparison to patients with BRCA mutation and control group. Higher tumor size (T) and lymph node metastases (N+) were negative prognostic factors independently from the presence of mutations and polymorphisms.

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