INHERITED MUTATIONS OF PALB2 GENE AND BREAST CANCER.

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Mutations in PALB2 predispose to breast cancer. There are two founder mutations of PALB2 in the Polish population (509_510delGA and 172_175delTTGT) which are associated with 4 to 5–fold increased risk of breast cancer. We found that 10-year survival for women with breast cancer and a PALB2 mutation is worse than of patients with breast cancer without a mutation (48% vs 75%, adjusted HR = 2.3; p<0·0001). Given that women with a PALB2 mutation face a high risk of breast cancer and are at a higher risk of death, increased surveillance should be offered to PALB2 carriers. It should be established whether unaffected PALB2 carriers benefit from prophylactic mastectomy, and if PABL2 carriers with breast cancer benefit from specific treatment, in particular specific chemotherapy regimens.