DO RECURRENT MUTATIONS IN GENES OTHER THAN BRCA1/2, CHEK2 AND PALB2 PLAY IMPORTANT ROLE IN PREDISPOSITION TO BREAST CANCER IN POLISH WOMEN?

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Approximately 20 genes other than BRCA1, BRCA2, CHEK2 and PALB2 have been associated with breast cancer predisposition, and extended genetic testing panels have been proposed. In Poland we found a number of founder mutations in BRCA1, CHEK2 and PALB2, however it is unknown if founder mutations in other candidate susceptibility genes play an important role in breast cancer in Polish women. Here we sought to establish if a single truncating mutation of XRCC2 (c.96delT, p.Phe32fs) detected in by whole-exome sequencing of 144 Polish women with familial breast cancer is recurrent mutation in Poland and whether it is associated with a genetic susceptibility to breast cancer in the population. We genotyped 3000 women with breast cancer and 2000 heathy women for the c.96delT mutation. The mutation was present with similar frequency in cases (0.23%) and in controls (0.25%). Our data suggest that c.96delT truncating mutation of XRCC2 is not associated with increased risk of breast cancer. This data calls into question adding of XRCC2 gene to breast cancer testing panels.