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Titel des Beitrags:
Germline mutations in the PALB2 gene are population specific and occur with low frequencies in familial breast cancer.

Abstract:
The Partner and Localizer of BRCA2 (PALB2) protein has been linked to Fanconi anemia and breast cancer predisposition. Here we present data of a comprehensive mutation screening of the PALB2 gene in 818 familial cases of breast cancer from Germany. By analyzing the entire coding region of PALB2, we found seven truncating mutations (six of them novel) in families tested negative for BRCA1/2-mutations. In addition, two novel potentially disease causing missense mutations were found. Remarkably, only one mutation reported previously in other populations, was also identified in the German population. No PALB2 mutation carriers were identified in 450 unaffected controls. Thus, our observations indicate a low prevalence of deleterious PALB2 mutations and a specific mutation profile within the German population. As PALB2-deficient tumors were shown to be sensitive to Poly(ADP-ribose) Polymerase (PARP) inhibitors, our study has implications for newly developed, favorable treatment options in familial breast cancer.

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