BRIP1 (BACH1) variants and familial breast cancer risk: a case-control study.

BACKGROUND: Inactivating and truncating mutations of the nuclear BRCA1-interacting protein 1 (BRIP1) have been shown to be the major cause of Fanconi anaemia and, due to subsequent alterations of BRCA1 function, predispose to breast cancer (BC). METHODS: We investigated the effect of BRIP1 -64G>A and Pro919Ser on familial BC risk by means of TaqMan allelic discrimination, analysing BRCA1/BRCA2 mutation-negative index patients of 571 German BC families and 712 control individuals. RESULTS: No significant differences in genotype frequencies between BC cases and controls for BRIP1 -64G>A and Pro919Ser were observed. CONCLUSION: We found no effect of the putatively functional BRIP1 variants -64G>A and Pro919Ser on the risk of familial BC.
TUM Einrichtung:
Frauenklinik und Poliklinik

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