Copy number variant in the candidate tumor suppressor gene MTUS1 and familial breast cancer risk.

Abstract:
Copy number variants (CNVs), insertions, deletions and duplications, contribute considerably to human genetic variation and disease development. A recent study has characterized 100 CNVs including a deletion in the mitochondrial tumor suppressor gene 1 (MTUS1) lacking the coding exon 4. MTUS1 maps to chromosome 8p, a region frequently deleted and associated with disease progression in human cancers, including breast cancer (BC). To investigate the effect of the MTUS1 CNV on familial BC risk, we analyzed 593 BC patients and 732 control individuals using a case-control study design. We found a significant association of the deletion variant with a decreased risk for both familial and high-risk familial BC (odds ratio (OR) = 0.58, 95% confidence interval (CI) = 0.37-0.90, P = 0.01 and OR = 0.41, 95% CI = 0.23-0.74, P = 0.003), supporting its role in human cancer. To our knowledge, the present study is the first to determine the impact of a CNV in a tumor suppressor gene on cancer risk.