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Keeping a CHEK on breast cancer

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Mutations in the tumor suppressor genes [BRCA1](#) and [BRCA2](#) account for a small percentage of breast cancer cases. In an Advanced Online Publication from [Nature Genetics](#), researchers from the CHEK-Breast Cancer Consortium report the identification of a mutation in the *CHEK2* gene that increases the risk of breast cancer in both women and men (22 April 2002, DOI: 10.1038/ng879). They performed a genome-wide linkage search in a family with *BRCA*-independent breast cancer and identified a mutation in the *CHEK2* gene that results in a truncated protein. [CHEK2](#) is a cell-cycle checkpoint kinase involved in DNA-damage-response pathways. The mutation abolished kinase activity, and is five times more frequent in families affected by breast cancer who do not have *BRCA1* or *BRCA2* mutations.

References

1. Genetic heterogeneity and penetrance analysis of the *BRCA1* and *BRCA2* genes in breast cancer families.
2. *Nature Genetics*, [<http://genetics.nature.com>]
3. The human homologs of checkpoint kinases Chk1 and Cds1 (Chk2) phosphorylate p53 at multiple DNA damage-inducible sites.