

Elucidation of the transmission of a novel mutation in BRCA1 (1125delCT) in a family with multiple cases of breast cancer.

[Razis E](#), [Belogianni I](#), [Siamanta V](#), [Apessos A](#), [Kyriakopoulou V](#), [Papadopoulou E](#), [Sariyanni M](#), [Theofilopoulos D](#), [Labropoulos S](#), [Nasioulas G](#).

Source

First Department of Medical Oncology, DTCA HYGEIA, 15123 Maroussi, Athens.

Abstract

In a family with multiple members affected by breast cancer we identified the novel mutation 1125delCT (exon 11) in BRCA1. Three out of three offsprings have the novel mutation while the mother affected by breast cancer does not carry the mutation. Linkage analysis revealed the transmission of the healthy haplotype from the mother to the three offsprings while the children inherited the mutated haplotype from the father. Our data document in an unquestionable way where the mutated haplotype was inherited from. In some families, although the transmission pathway seems obvious, the molecular analysis yields surprising results.