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We have analyzed a single multi-affected breast/ovarian cancer pedigree (BOV3) and have shown consistent inheritance of markers on chromosome 17q with the disease confirming that this family is due to the BRCA1 gene. Analysis of 17q haplotypes shows a recombination event in a bilateral breast cancer case which suggests that the BRCA1 gene lies distal to D17S857; D17S857 is thus the new proximal boundary for the region containing BRCA1. Combining this information with previously published mapping information suggests that BRCA1 is contained in a region estimated at 1-1.5 Mb in length. All seven breast tumour/blood pairs examined from this family show loss of heterozygosity in the tumours. The allele retained in each tumour was from the disease-bearing chromosome implicating BRCA1 as a tumour suppressor gene. We have sequenced the 17 beta-oestradiol dehydrogenase genes (EDH17B1 and EDH17B2) which have been suggested as candidate genes for BRCA1 in four members of this family. No germline mutations were detected.

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