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Breast Cancer Res Treat. 1993 Nov;28(2):121-35.**Molecular cloning of BRCA1: a gene for early onset familial breast and ovarian cancer.**Bowcock AM.**Author information****Abstract**

Molecular analyses allow one to determine genetic lesions occurring early in the development of tumors. With positional cloning approaches we are searching for a gene involved in the development of early onset familial breast and ovarian cancer that maps to human chromosome 17q21 and is termed **BRCA1**. This involves localizing the region genetically within families with multiply affected members, capturing the region identified by genetic analyses in YACs (yeast artificial chromosomes), converting those YACs to smaller manipulable pieces (such as cosmids), and searching for **genes** via a variety of approaches such as direct screening of cDNA libraries with genomic clones, direct selection by hybridization, "exon trapping", and CpG island rescue. Once identified, candidate **genes** will be screened for mutations in affected family members in whom breast cancer segregates with the locus on 17q21. The frequency of this gene has been calculated to be 0.0033; from this the incidence of carriers, i.e. those carrying such a predisposition, is one in 150 women. The isolation of **BRCA1** and the elucidation of the mutations resulting in breast and ovarian cancer predisposition will allow identification of women who have inherited germ-line mutations in **BRCA1**. In families known to harbor a germ-line **BRCA1** mutation, diagnosis of affected members will be rapid. It is possible that one will also be able to detect alterations of the second copy of this gene early in tumor development in individuals carrying a germ-line mutation. It is not yet known how frequently somatic **BRCA1** mutations predispose to breast and ovarian carcinoma in the general female population. If, as in other genetic diseases, new germ-line mutations occur in some women and thus contribute to the development of breast cancer, it may be feasible to screen women in the general population for predisposing mutations. In addition, if acquired genetic mutations of the **BRCA1** gene are involved as early events in the development of non-familial forms of the disease, early detection of possible breast carcinoma may become feasible in biopsy of breast tissue.

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