

BRCA1

BRCA1 (*Breast cancer 1, early onset*) is a tumour suppressor gene. The protein encoded by this gene is important in DNA repair. The DNA damage may be caused by natural or medical irradiation, or other environmental exposures, and may also occur during cell division. *BRCA1* is involved in the response to double-stranded DNA breaks, both in sensing damage and in coordinating its repair.

The *BRCA1* gene is located on the long arm of chromosome 17, at position 21 (17q21)

Over 1000 mutations in the *BRCA1* gene have been identified, many of which are associated with an increased incidence of breast cancer. A defective or missing *BRCA1* protein cannot repair damaged DNA or fix mutations in other genes, ultimately leading to tumour formation.

BRCA1 is strongly associated with breast cancer in women, but is also associated with an increased incidence in men. In addition other cancers, such as ovarian cancer, are more common in women with a mutation in the *BRCA1* gene.

Nomenclature dictates that the gene itself is written in italics, while its protein product is not.

BRCA2

BRCA2 is also a tumour suppressor gene involved in DNA repair. It functions in conjunction with a protein encoded by *PALB2*, which co-localises with *BRCA2* in the nucleus. Recent evidence suggests that mutations of *PALB2*, like *BRCA2*, are associated with an increased risk of breast cancer and ovarian cancer, as well as pancreatic cancer, prostate cancer and malignant melanoma.

BRCA2 is located on chromosome 13.