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**BRCA1**



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*Reviewed August 2007*

## What is the official name of the *BRCA1* gene?

The official name of this gene is “breast cancer 1, early onset.”

*BRCA1* is the gene's official symbol. The *BRCA1* gene is also known by other names, listed below.

Read more about gene names and symbols on the [About](#) page.

## What is the normal function of the *BRCA1* gene?

The *BRCA1* gene belongs to a class of genes known as tumor suppressor genes. Like many other tumor suppressors, the protein produced from the *BRCA1* gene helps prevent cells from growing and dividing too rapidly or in an uncontrolled way.

The *BRCA1* gene provides instructions for making a protein that is directly involved in repairing damaged DNA. In the nucleus of many types of normal cells, the BRCA1 protein interacts with several other proteins, including the proteins produced from the *RAD51* and *BARD1* genes, to mend breaks in DNA. These breaks can be caused by natural and medical radiation or other environmental exposures, and also occur when chromosomes exchange genetic material in preparation for cell division. By helping repair DNA, BRCA1 plays a role in maintaining the stability of a cell's genetic information.

Research suggests that the BRCA1 protein also regulates the activity of other genes and plays a critical role in embryonic development. To carry out these functions, the BRCA1 protein interacts with many other proteins, including other tumor suppressors and proteins that regulate cell division.

## Does the *BRCA1* gene share characteristics with other genes?

The *BRCA1* gene belongs to a family of genes called [RNF](#) (RING-type zinc fingers).

A gene family is a group of genes that share important characteristics. Classifying individual genes into families helps researchers describe how genes are related to each other. For more information, see [What are gene families?](#) in the Handbook.

## How are changes in the *BRCA1* gene related to health conditions?

[breast cancer](#) - increased risk from variations of the *BRCA1* gene

Researchers have identified more than 1,000 mutations in the *BRCA1* gene, many of which are associated with an increased risk of cancer (particularly breast cancer in women). Most of these mutations lead to the production of an abnormally short version of the BRCA1 protein, or prevent any protein from being made from one

copy of the gene. Other *BRCA1* mutations change single protein building blocks (amino acids) in the protein or delete large segments of DNA from the *BRCA1* gene. Researchers believe that a defective or missing BRCA1 protein is unable to help repair damaged DNA or fix mutations that occur in other genes. As these defects accumulate, they can allow cells to grow and divide uncontrollably and form a tumor.

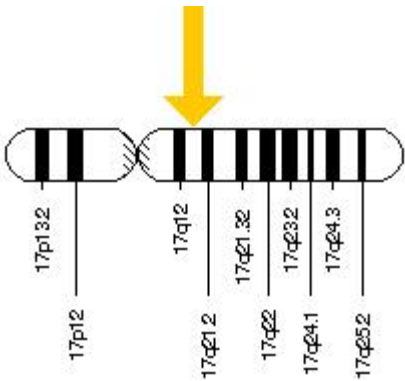
other cancers - increased risk from variations of the *BRCA1* gene

In addition to female breast cancer, mutations in the *BRCA1* gene also increase the risk of several other types of cancer including fallopian tube cancer, male breast cancer, and pancreatic cancer. Many of these mutations change one of the amino acids used to make the BRCA1 protein, resulting in a protein that cannot perform its normal DNA repair function.

**Where is the *BRCA1* gene located?**

Cytogenetic Location: 17q21

Molecular Location on chromosome 17: base pairs 43,044,294 to 43,125,482



The *BRCA1* gene is located on the long (q) arm of [chromosome 17](#) at position 21.

More precisely, the *BRCA1* gene is located from base pair 43,044,294 to base pair 43,125,482 on chromosome 17.

See [How do geneticists indicate the location of a gene?](#) in the Handbook.

**Where can I find additional information about *BRCA1*?**

You and your healthcare professional may find the following resources about *BRCA1* helpful.

- [Educational resources](#) - Information pages (2 links)
- [Gene Reviews](#) - Clinical summary
- [Genetic Testing Registry](#) - Repository of genetic test information (1 link)

You may also be interested in these resources, which are designed for genetics professionals and researchers.

- [PubMed](#) - Recent literature
- [OMIM](#) - Genetic disorder catalog
- [Research Resources](#) - Tools for researchers (5 links)

## What other names do people use for the *BRCA1* gene or gene products?

- BRCA1 gene
- BRCA1\_HUMAN
- BRCC1
- Breast Cancer 1, Early Onset Gene
- Breast Cancer 1 Gene
- Breast Cancer, Type 1
- Breast Cancer Type 1 Susceptibility Gene
- Breast cancer type 1 susceptibility protein
- breast-ovarian cancer, included
- Hereditary Breast Cancer 1
- IRIS
- PPP1R53
- PSCP
- RNF53

See [How are genetic conditions and genes named?](#) in the Handbook.

## Where can I find general information about genes?

The Handbook provides basic information about genetics in clear language.

- [What is DNA?](#)
- [What is a gene?](#)
- [How do genes direct the production of proteins?](#)
- [How can gene mutations affect health and development?](#)

These links provide additional genetics resources that may be useful.

- [Genetics education](#)
- [Human Genome Project](#)
- [Resources for Genetics Researchers](#)

## What glossary definitions help with understanding *BRCA1*?

[acids](#) ; [cancer](#) ; [cell](#) ; [cell division](#) ; [class](#) ; [DNA](#) ; [DNA repair](#) ; [embryonic](#) ; [gene](#) ; [nucleus](#) ; [ovarian](#) ; [pancreatic](#) ; [protein](#) ; [radiation](#) ; [susceptibility](#) ; [susceptibility gene](#) ; [tumor](#)

You may find definitions for these and many other terms in the Genetics Home Reference [Glossary](#).

See also [Understanding Medical Terminology](#).

## [References](#) (18 links)

The resources on this site should not be used as a substitute for professional medical care or advice. Users seeking information about a personal genetic disease, syndrome, or condition should consult with a qualified healthcare professional. See [How can I find a genetics professional in my area?](#) in the Handbook.

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