BRCA2 gene

BRCA2 DNA repair associated

Normal Function

The *BRCA2* gene provides instructions for making a protein that acts as a tumor suppressor. Tumor suppressor proteins help prevent cells from growing and dividing too rapidly or in an uncontrolled way.

The BRCA2 protein is involved in repairing damaged DNA. In the nucleus of many types of normal cells, the BRCA2 protein interacts with several other proteins to mend breaks in DNA. These breaks can be caused by natural and medical radiation or other environmental exposures, and they also occur when chromosomes exchange genetic material in preparation for cell division. By helping to repair DNA, the BRCA2 protein plays a critical role in maintaining the stability of a cell's genetic information.

Researchers suspect that the BRCA2 protein has additional functions within cells. For example, the protein may help regulate cytokinesis, which is the step in cell division when the fluid surrounding the nucleus (the cytoplasm) divides to form two separate cells. Researchers are investigating the protein's other potential activities.

Health Conditions Related to Genetic Changes

Breast cancer

Mutations in the *BRCA2* gene are associated with an increased risk of breast cancer in both men and women, as well as several other types of cancer. These mutations are present in every cell in the body and can be passed from one generation to the next. As a result, they are associated with cancers that cluster in families. However, not everyone who inherits a mutation in the *BRCA2* gene will develop cancer. Other genetic, environmental, and lifestyle factors also contribute to a person's cancer risk.

Most *BRCA2* gene mutations lead to the production of an abnormally small, nonfunctional version of the BRCA2 protein from one copy of the gene in each cell. As a result, less of this protein is available to help repair damaged DNA or fix mutations that occur in other genes. As these defects accumulate, they can trigger cells to grow and divide uncontrollably to form a tumor.

Ovarian cancer

Many of the same *BRCA2* gene mutations that increase the risk of breast cancer (described above) also increase the risk of ovarian cancer. Families with these mutations are often said to be affected by hereditary breast and ovarian cancer syndrome. Women with *BRCA2* gene mutations have an approximately 12 to 25 percent chance of developing ovarian cancer in their lifetimes, as compared with 1.6 percent in the general population.

Prostate cancer

Inherited *BRCA2* gene mutations have been found to increase the risk of prostate cancer. Men with these mutations are also more likely to develop prostate cancer at an earlier age and may be at increased risk of having an aggressive form of the disease. They may also be at increased risk for other cancers.

BRCA2 gene mutations likely reduce the BRCA2 protein's ability to repair DNA, allowing potentially damaging mutations to persist in various other genes. The accumulation of damaging mutations can lead to the out-of-control cell growth and division that can result in development of a tumor.

Cholangiocarcinoma

MedlinePlus Genetics provides information about Cholangiocarcinoma

Fanconi anemia

MedlinePlus Genetics provides information about Fanconi anemia

Other cancers

Inherited mutations in the *BRCA2* gene also increase the risk of several other types of cancer, including pancreatic cancer and an aggressive form of skin cancer called melanoma. These mutations impair the ability of the BRCA2 protein to help repair damaged DNA. As defects accumulate in DNA, they can trigger cells to grow and divide without order to form a tumor. It is not clear why different individuals with *BRCA2* mutations develop cancers in different organs. Environmental factors that affect specific organs may contribute to the development of cancers at particular sites.

Other Names for This Gene

- brca 2 gene
- BRCA2_HUMAN
- BRCC2
- breast cancer 2
- breast cancer 2 gene
- breast cancer 2, early onset
- breast cancer 2, early onset gene

- breast cancer type 2 susceptibility gene
- breast cancer type 2 susceptibility protein
- FACD
- FAD
- FAD1
- FANCB
- FANCD1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of BRCA2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=675[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28BRCA2%5BMAJR%5D%29+AND+%28BRCA2%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- BRCA2 DNA REPAIR-ASSOCIATED PROTEIN; BRCA2 (https://omim.org/entry/600185)
- FANCONI ANEMIA, COMPLEMENTATION GROUP D1; FANCD1 (https://omim.org/entry/605724)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/675)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=BRCA2[gene])

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Genomic Location

The *BRCA2* gene is found on chromosome 13 (https://medlineplus.gov/genetics/chromosome/13/).

Last updated May 1, 2020