BRCA1 Gene

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1. Normal Function

The *BRCA1* 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 BRCA1 protein is involved in repairing damaged DNA. In the nucleus of many types of normal cells, the BRCA1 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 BRCA1 protein plays a critical 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 an essential 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.

2. Health Conditions Related to Genetic Changes

2.1. Breast Cancer

Mutations in the *BRCA1* 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 *BRCA1* gene will develop cancer. Other genetic, environmental, and lifestyle factors also contribute to a person's cancer risk.

Most *BRCA1* gene 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. 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.

2.2. Ovarian Cancer

Many of the same *BRCA1* 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 *BRCA1* gene mutations have a 35 to 60 percent chance of developing ovarian cancer in their lifetimes, as compared with 1.6 percent in the general population.

2.3. Prostate Cancer

Inherited *BRCA1* gene mutations have been found to increase the risk of prostate cancer. These mutations likely reduce the BRCA1 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 cause a tumor to develop. Men who carry a *BRCA1* gene mutation may also be at increased risk for other cancers, including breast and pancreatic cancer.

2.4. Cholangiocarcinoma

Cholangiocarcinoma

2.5. Other Cancers

Inherited mutations in the *BRCA1* gene also increase the risk of several other types of cancer, including pancreatic cancer and colon cancer. These mutations impair the ability of the BRCA1 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 *BRCA1* mutations develop cancers in different organs. Environmental factors that affect specific organs may contribute to the development of cancers at particular sites.

3. Other Names for This Gene

- BRCA1 gene
- BRCA1_HUMAN
- BRCC1
- breast cancer 1
- breast cancer 1 gene
- breast cancer 1, early onset
- breast cancer 1, early onset gene
- breast cancer type 1 susceptibility gene
- breast cancer type 1 susceptibility protein
- IRIS
- PPP1R53
- PSCP
- RNF53

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