

The Genetics of Ovarian Cancer

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What are genes?

Genes carry information in the form of DNA within each cell of the human body. Researchers estimate that there are 30,000 different genes in each cell. Genes are packaged onto chromosomes. There are 23 pairs of chromosomes in each cell. One chromosome of each pair is inherited from the person's father and one from the person's mother.

Genes control how a cell functions, including how quickly it grows, how often it divides, and how long it lives. To control these functions, genes produce proteins that perform specific tasks and act as messengers for the cell. Therefore, it is essential that each gene have the correct instructions or "code" for making its protein so that the protein can perform the proper function for the cell.

What role do genes play in ovarian cancer?

Many cancers begin when one or more genes in a cell are mutated (changed), creating an abnormal protein or no protein at all. The information provided by an abnormal protein is different from that of a normal protein, which can cause cells to multiply uncontrollably and become cancerous.

A person may either be born with the genetic mutation in all of their cells (germline mutation) or acquire a genetic mutation in a single cell during his or her lifetime. An acquired mutation is passed on to all cells that develop from that single cell (called a somatic mutation). Somatic mutations can sometimes be caused by environmental factors, such as cigarette smoke. Most ovarian cancers (about 85% to 90%) are considered sporadic, meaning that the damage to the genes occurs by chance after a person is born and there is no risk of passing on the gene to a person's children. Inherited ovarian cancers are less common (about 10% to 15%) and occur when gene mutations are passed within a family, from one generation to the next.

What are the chances a mutated gene is inherited?

Every cell usually has two copies of each gene: one inherited from a person's mother and one inherited from a person's father. Most types of hereditary ovarian cancer follow an autosomal dominant inheritance pattern, in which a mutation needs to happen in only one copy of the gene for the person to have an increased risk of getting the disease. This means that a parent with a gene mutation may pass on a copy of the normal gene or a copy of the gene with a mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a gene mutation also has a 50% chance of having the same mutation.

What is a woman's average risk for ovarian cancer?

A woman's average risk of developing ovarian cancer during her lifetime is about one in 71.

How does a woman know if she has inherited a genetic mutation that increases her risk of ovarian cancer?

Only [genetic testing \[2\]](#) can determine whether a person has a genetic mutation. Most experts strongly recommend that people considering genetic testing first consult a [genetic counselor \[3\]](#). Genetic counselors are trained to explain the risks and benefits of genetic testing.

How does a woman know if ovarian cancer runs in her family?

Ovarian cancer may run in the family if first-degree relatives (mother, sisters, daughters) or many other family members (grandmothers, aunts, nieces, granddaughters) have had ovarian cancer.

What is a woman's risk if ovarian cancer runs in her family?

If a woman's first-degree relatives developed ovarian cancer, her risk of ovarian cancer is about three times higher than the average woman's risk of ovarian cancer. The risk increases if other close relatives have had ovarian cancer.

Which inherited genetic mutations raise the risk of ovarian cancer?

There are several genetic conditions linked to an increased risk of ovarian cancer. Some of the most common hereditary cancer syndromes associated with ovarian cancer risk are described below.

[Hereditary breast and ovarian cancer \(HBOC\) syndrome \[4\]](#). HBOC is associated with mutations in the *BRCA1* and/or *BRCA2* (*BRCA* stands

for BREast CAncer). Women with HBOC have an increased risk of [breast cancer](#) [5] and ovarian cancer.

Lynch syndrome [6]. Lynch syndrome increases a woman's risk of ovarian and [uterine cancer](#) [7]. It is caused by mutations in several different genes. Lynch syndrome also increases the risk of [colorectal cancer](#) [8], as well as cancers of the [stomach](#) [9], [small intestine](#) [10], [liver](#) [11], [bile duct](#) [12], [urinary tract](#) [13], the [brain and central nervous system](#) [14], and possibly [breast cancer](#) [5].

Peutz-Jeghers syndrome (PJS) [15]. PJS is caused by a specific genetic mutation and is associated with multiple polyps in the digestive tract that become noncancerous tumors, increased pigmentation (dark spots on the skin) on the face and hands. In addition to an increased risk of ovarian cancer, PJS also raises risk of [breast cancer](#) [5], [uterine](#) [7], and [lung](#) [16] cancers.

Nevoid basal cell carcinoma syndrome (NBCCS) [17]. Women with NBCCS (also called Gorlin syndrome) have an increased risk of developing fibromas (benign fibrous tumors) of the ovaries. There is a small risk that these fibromas could develop into a type of ovarian cancer called fibrosarcoma. People with NBCCS often have multiple [basal cell carcinomas](#) [18] and jaw cysts and may develop [medulloblastoma](#) [19] (a type of brain tumor) in childhood.

Are there other genetic conditions associated with an increased risk of ovarian cancer?

Other genetic conditions that are associated with a small increased risk of ovarian cancer are described below.

Li-Fraumeni syndrome (LFS) [20]. LFS is a rare condition associated with a specific genetic mutation. People with LFS have a higher risk of developing [osteosarcoma](#) [21] (a type of bone cancer), [soft tissue sarcoma](#) [22], [leukemia](#) [23], [breast cancer](#) [5], [brain cancer](#) [14], and [adrenal cortical tumors](#) [24].

Ataxia telangiectasia (A-T) [25]. A-T is a rare disorder associated with a specific genetic mutation. It causes progressive neurological problems that lead to difficulty walking, slurred speech, and difficulty with writing and other tasks. People with A-T have an increased risk of leukemia and lymphoma, and possibly [sarcoma](#) [22], [breast cancer](#) [5], ovarian cancer, and [stomach cancer](#) [9].

What is your risk level?

In addition to family history, other environmental and lifestyle factors may increase your risk of ovarian cancer. Discussing your family history and personal risk factors with a doctor helps you better understand your risk. People with a higher than average risk may benefit from genetic counseling and early detection strategies. Some women choose to have a prophylactic oophorectomy (the preventive removal of healthy ovaries), which reduces the risk of both breast and ovarian cancers. Prophylactic surgery does not, however, completely eliminate the risk of either breast or ovarian cancer.

A [risk factor](#) [26] is anything that increases a person's risk of developing cancer. Having a particular genetic mutation linked to ovarian cancer cannot predict that a person will develop cancer. Controllable risk factors, such as eating a balanced diet, maintaining a healthy weight, exercising, limiting alcoholic beverages, and avoiding tobacco products also play a role. Most women who develop ovarian cancer have few known risk factors. Research to better understand the link between genetic mutations and ovarian cancer is ongoing. Talk with a doctor for more information about risk factors, prevention, and screening for ovarian cancer.

More Information

[Genetics](#) [27]

[Guide to Ovarian Cancer](#) [28]

[Sharing Genetic Test Results With Your Family](#) [29]

[Direct-to-Consumer Genetic Testing](#) [30]

[Cancer Advances: Extent of Cancer Risk Reduction Depends Through Ovary Removal Depends on BRCA Mutation Type](#) [31]

Links:

[1] <http://www.cancer.net/about-us>

[2] <http://www.cancer.net/patient/All+About+Cancer/Genetics/Genetic+Testing>

[3] <http://www.cancer.net/patient/All+About+Cancer/Genetics/What+to+Expect+When+Meeting+With+a+Genetic+Counselor>

[4] <http://www.cancer.net/patient/Cancer+Types/Hereditary+Breast+and+Ovarian+Cancer>

[5] <http://www.cancer.net/patient/Cancer+Types/Breast+Cancer>

[6] <http://www.cancer.net/patient/Cancer+Types/Lynch+Syndrome>

[7] <http://www.cancer.net/patient/Cancer+Types/Uterine+Cancer>

[8] <http://www.cancer.net/patient/Cancer+Types/Colorectal+Cancer>

[9] <http://www.cancer.net/patient/Cancer+Types/Stomach+Cancer>

[10] <http://www.cancer.net/patient/Cancer+Types/Small+Bowel+Cancer>

[11] <http://www.cancer.net/patient/Cancer+Types/Liver+Cancer>
[12] <http://www.cancer.net/patient/Cancer+Types/Bile+Duct+Cancer>
[13] <http://www.cancer.net/patient/Cancer+Types/Bladder+Cancer>
[14] <http://www.cancer.net/patient/Cancer+Types/Brain+Tumor>
[15] <http://www.cancer.net/patient/Cancer+Types/Peutz-Jeghers+Syndrome>
[16] <http://www.cancer.net/patient/Cancer+Types/Lung+Cancer>
[17] <http://www.cancer.net/patient/Cancer+Types/Nevoid+Basal+Cell+Carcinoma+Syndrome>
[18] <http://www.cancer.net/patient/Cancer+Types/Skin+Cancer+%28Non-Melanoma%29>
[19] <http://www.cancer.net/patient/Cancer+Types/Medulloblastoma+-+Childhood>
[20] <http://www.cancer.net/patient/Cancer+Types/Li-Fraumeni+Syndrome>
[21] <http://www.cancer.net/patient/Cancer+Types/Osteosarcoma+-+Childhood>
[22] <http://www.cancer.net/patient/Cancer+Types/Sarcoma>
[23] <http://www.cancer.net/patient/Cancer+Types>
[24] <http://www.cancer.net/patient/Cancer+Types/Adrenal+Gland+Tumor>
[25] <http://www.cancer.net/patient/Cancer+Types/Ataxia-Telangiectasia>
[26] <http://www.cancer.net/patient/All+About+Cancer/Risk+Factors+and+Prevention>
[27] <http://www.cancer.net/patient/All+About+Cancer/Genetics>
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[29] <http://www.cancer.net/patient/All+About+Cancer/Genetics/Sharing+Genetic+Test+Results+With+Your+Family/Sharing+Genetic+Test+Results+With+Your+Family>
[30] <http://www.cancer.net/patient/All+About+Cancer/Cancer.Net+Feature+Articles/Cancer+Screening+and+Prevention/ASCO+Expert+Corner%3A+Direct-to-Consumer+Genetic+Testing>
[31] <http://www.cancer.net/patient/Publications+and+Resources/Cancer+Advances/News+for+Patients+from+the+Journal+of+Clinical+Oncology/Cancer+Advances%3A+Extent+of+Cancer+Risk+Reduction+through+Ovary+>