Yale NewHaven Health
Smilow Cancer Hospital

Yale CANCER CENTER
A Comprehensive Cancer Center Designated by the National Cancer Institute

Hereditary ovarian cancer

Smilow Cancer Genetics and Prevention Program

While most ovarian cancer is not hereditary, an estimated 10-15 percent of ovarian cancer is due to an inherited cause. This means that the cancer runs in the family.

Finding a hereditary explanation for why you have ovarian cancer can help to:

- Guide the course of your cancer treatment or any decisions about surgery
- Alert you and your providers about possible risks for other types of cancers

What are the risks?

Most cases of hereditary ovarian cancer are caused by inherited mutations in two genes, called *BRCA1* and *BRCA2.* Women who have a BRCA1 or BRCA2 inherited gene mutation have an increased risk of ovarian cancer, including cancer of the fallopian tubes and peritoneum. Estimates of these risks vary greatly, ranging from 13 to 58 percent. By age 70, women who have a BRCA1 or BRCA2 inherited gene mutation also have a 60 percent chance of developing breast cancer. In the general population of women who do not have an inherited mutation, the risk of getting ovarian cancer during her lifetime is about two percent for ovarian cancer and approximately 12 percent for breast cancer.

Men with mutations in the BRCA1 and BRCA2 genes have a seven percent risk of developing breast cancer and up to a 61 percent of getting prostate cancer. In the general population, the lifetime risks for male breast cancer are less than one percent and 12 percent for prostate cancer is 12 percent. Both men and women with inherited mutations in BRCA1/2 genes have a nearly eight percent risk of pancreatic cancer and an increased risk for melanoma. This is particularly the case for those with a BRCA2 mutation.

The importance of genetic testing

Genetic testing is one way to find out if you have a hereditary risk for ovarian cancer. Genetic testing can look for several hereditary cancer syndromes by testing a panel of genes (including BRCA1 and BRCA2) that are related to hereditary ovarian cancer. The risk for ovarian and other cancers associated with mutations in these genes can vary (see tables on next pages for details). It is important to remember that some of the genes tested on the panel have not been studied as long as other genes, and that some genes only have a possible association with ovarian cancer risk. For these less-studied genes, information about associated cancer risks and any screening recommendations may change over time.

It is also important to share the results of genetic testing with relatives because they may have also inherited the same increased cancer risks. A parent who has a mutation in one of these ovarian cancer genes has a 50 percent chance of passing the mutation on to each of his or her children. It also means his or her siblings have a 50 percent chance that they have the mutation as well. When a hereditary explanation is found in a family, relatives can also have genetic testing to better understand their risks. This will help them guide their own decisions about cancer screening, prevention and management.

Hereditary Cancer Syndrome and Gene(s)	Lifetime Risk of Ovarian Cancer	Increased Risk for Other Cancers
Hereditary Breast and Ovarian Cancer **Genes: *BRCA1, BRCA2***	13-58 percent	- Breast cancer - Pancreatic cancer - Prostate cancer - Melanoma
Gene: *BRIP1*	5-15 percent	- May have elevated risk for breast cancer, particularly triple negative-type
Lynch Syndrome **Genes: *MLH1, MSH2, MSH6, PMS2, EPCAM***	3-20 percent Risks varies by gene	- Colon and other gastrointestinal cancers - Uterine cancer - Urinary tract cancer - Sebaceous skin cancer
Gene: *RAD51C, RAD51D*	10-20 percent	- Elevated risk for breast cancer, particularly triple negative-type
Li-Fraumeni Syndrome **Gene: *TP53***	Possibly increased, but not well defined	- Female breast cancer - Risk to develop multiple different types of cancer
Gene: *ATM*	Up to 3 percent	- Female breast cancer - Pancreatic cancer
Gene: *PALB2*	Up to 5 percent	- Female breast cancer - Pancreatic cancer

Syndromes Associated with Less Common Types of Ovarian Cancer	Lifetime Risk of Ovarian Cancer	Increased Risk for Other Cancers
Peutz-Jeghers syndrome **Genes: *STK11***	Up to 21 percent	- Gastrointestinal (GI) tract polyps - Colorectal, GI and pancreatic cancer - Breast cancer - Uterine and a rare type of cervical cancer
DICER1* Tumor Predisposition syndrome** **Gene: *DICER1	Increased, but not well defined	- Tumors of the lungs, kidneys and thyroid mostly presenting in childhood - Other more rare tumors can be seen - Polyps of the small intestine

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