Health Enews

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Genetic Testing for Breast & Ovarian Cancer

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BRCA1 and BRCA2 gene mutation testing is recommended in people who might be at risk for BRCA gene mutation. Candidates include:

- Breast cancer diagnosed before the age of 50
- Cancer in both breasts in the same woman
- Both breast and ovarian cancers in either the same woman or the same family
- Multiple breast cancers
- Two or more primary types of BRCA1 or BRCA2 related cancers in a single family member
- Cases of male breast cancer
- Ashkenazi Jewish ethnicity

Other Cancers Linked to BRCA1 and BRCA2 Mutations

- Increased risk of developing fallopian and peritoneal cancer in women.
- High risk of prostate cancer in men.
- Increased risk of pancreatic cancer in men and women.
- Increased risk of Fanconi anemia subtype (FA-D1), a syndrome that is associated with childhood solid tumors and development of acute myeloid leukemia.

How to Prepare for BRCA Gene Testing

- Meet with a genetic counselor who will determine whether it's appropriate for you and discuss the potential risks, limitations, and benefits.
- Gather information about your family's medical history, especially that of close relatives.
- Write down questions for the counselor.
- Try to prepare yourself for the emotional and social implications that learning your genetic status might have. Test results could also fail to provide you with clear-cut answers regarding your cancer risk, so prepare to face that possibility also.

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What you can expect

- The BRCA gene test is most often a blood test. Occasionally, saliva and skin biopsies are also needed.
- It may take several weeks before test results are available.
- When results are available, you will meet with a genetic counselor to learn your results and discuss any possible implications.

Results

If your test is positive:

- A positive test result indicates that a person has inherited a known harmful mutation in BRCA1 or BRCA2 and, therefore, has an increased risk of developing certain cancers. However, a positive test result cannot tell whether or when an individual will actually develop cancer. For example, some women who inherit a harmful BRCA1 or BRCA2 mutation will never develop breast or ovarian cancer.
- Experts recommend that women who carry the BRCA1 or BRCA2 mutation undergo clinical breast examinations beginning at age 25 to 35 years. Additionally, a mammogram should be completed every year beginning at age 25 to 35 years of age.
- The American Cancer Society and the National Comprehensive Cancer Network, recommend that annual screenings with mammography *and* MRI for women who have a high risk for breast cancer. An MRI may be more sensitive than mammography for women at high risk of breast cancer.
- Women with a positive test result may choose to undergo a bilateral prophylactic mastectomy to reduce their risk of breast cancer and a bilateral prophylactic salpingo-oophorectomy to reduce the risk of developing ovarian cancer. Research has shown that women who underwent a salpingo-oophorectomy had a nearly 80% reduction in dying from ovarian cancer and a 56% reduction in risk of dying from breast cancer.
- Unfortunately, there is no known effective ovarian cancer screening that currently exists. Some groups recommend ultrasound, clinical examination, and a blood test for antigen CA-125.
- Additionally, it is suggested that men who carry the BRCA1 or BRCA2 mutation undergo regular mammography as well as testing for prostate cancer. However, the value of these screenings remain unproven at present.

If your test is negative:

- A negative test result can be more difficult to understand than a positive result because the result depends on family history of cancer and whether a BRCA1 or BRCA2 mutation was identified in the blood relative with cancer.
- If a first or second degree relative is known to carry the BRCA1 or BRCA2 mutation, and your results are negative, than a negative result is clearly a true negative.

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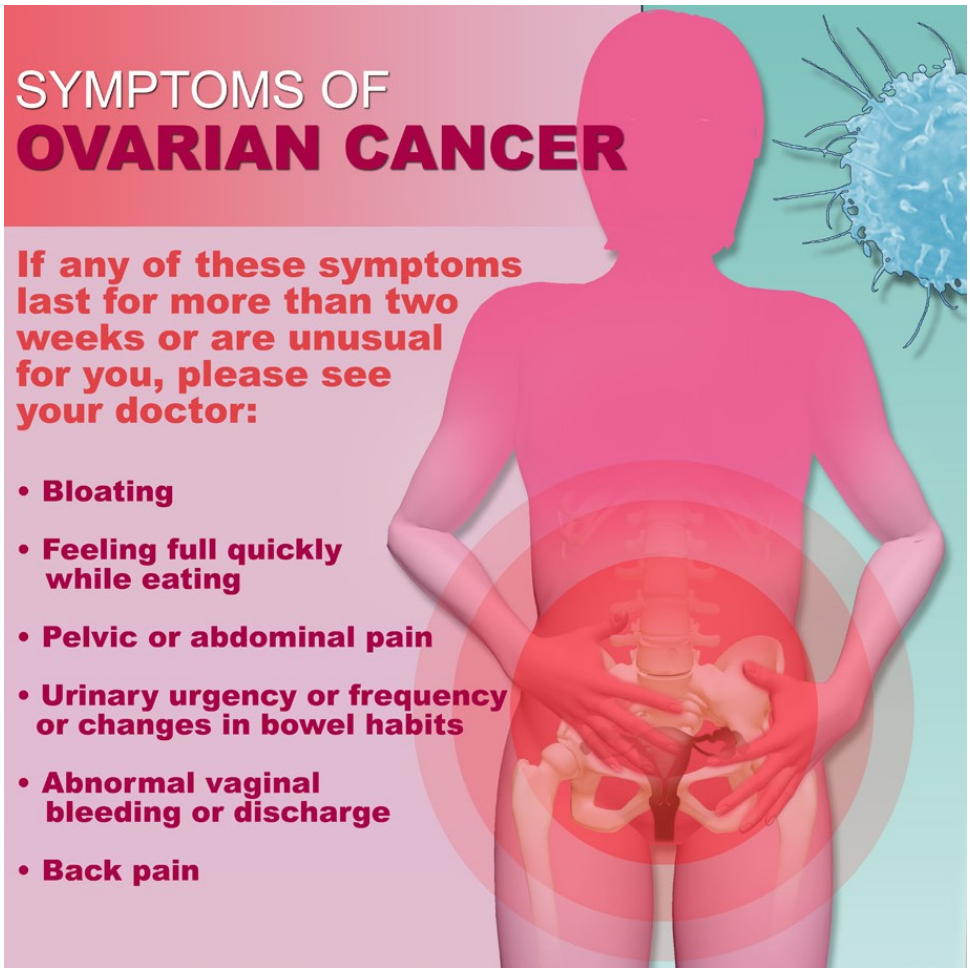
If your test is ambiguous or uncertain:

- Genetic testing can discover a change in BRCA1 or BRCA2 that has not previously been associated with cancer. The test results are considered ambiguous because it isn't known whether this specific gene change can affect a person's risk of developing cancer. Over time, additional studies of variants of uncertain significance may result in a specific mutation being reclassified as either harmful or clearly not harmful.

Insurance Laws and BRCA1 and BRCA2 Genetic Mutation Testing:

- The Affordable Care Act considers genetic counseling and BRCA1 and BRCA2 mutation testing for individuals at high risk a covered preventative service.
- Federal and state laws help ensure the privacy of your genetic information and protect against discrimination in health insurance and employment.



http://www.mayoclinic.org/tests-procedures/brca-gene-test/basics/what-you-can-expect/prc-20020361
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