
Genetic Services Policy Project

Inherited Breast and Ovarian Cancers

What are Inherited Breast and Ovarian Cancers (BOC)?

Inherited BOC are autosomal dominant conditions with reduced penetrance and associated with a high lifetime risk of breast and ovarian cancer.

What is known about the genetic and non-genetic causes of inherited BOC?

- Most BOC are sporadic and occur among people without family history, but heredity may account for 5 to 10% of BOC diagnosed each year in the United States.
- Inherited BOC may involve mutations in the BRCA1 and BRCA2 genes. Prevalence of BRCA mutations is estimated to be 1 in 500. Other genes may also contribute to inherited BOC.
- Family history of inherited BOC may represent:
 - genetic causes of BOC
 - shared environmental causes of BOC
 - both genetic causes and shared environmental causes of BOC.

How are inherited BOC detected?

- Families with inherited BOC are often identified by family history of cancer. Key elements of family history include:
 - Two first degree relatives with breast cancer, one with a diagnosis before age 50
 - Three or more first or second degree relatives, regardless of age of onset
 - Both breast and ovarian cancer among first and second degree relatives
 - First degree relative with bilateral breast cancer
 - Two or more first or second degree relatives with ovarian cancer
 - First or second degree relative with both breast and ovarian cancer
 - Male relative with breast cancer
- Many different types of clinical providers may help to identify family history for BOC.
- Cancer screening is often used for early cancer detection.
 - Breast cancer screening for persons at high risk: initiated by age 30, with mammography and MRI screening used alternately on a 6-monthly basis.
 - Ovarian cancer screening: vaginal ultrasound and measurement of serum CA-125 level
- Genetic testing for BRCA mutations has been available since 1996.

What are preventive therapies for BOC?

- Tamoxifen: women with a BRCA2 mutation may benefit from this treatment, but women with a BRCA1 mutation may not.
- Risk-reducing mastectomy.
- Risk-reducing oophorectomy.

What are costs associated with BOC?

- There are no cost estimates related to inherited BOC. However, studies using Medicare expenditure data have provided estimates for costs of metastatic breast cancer, which may include inherited breast cancer. Mean total costs for a patient with metastatic breast cancer during a 16 month time period were \$35,164 compared to \$4,176 for a person without metastatic breast cancer.
- Total annual medical care treatment costs for women 45 and older with breast and all gynecological cancers from all causes average \$5.0 billion.

References

- Easton, D., D. Ford, et al. (1993). "Inherited susceptibility to breast cancer." Cancer Surv 18: 95-113.
- National Cancer Institute. (2006, January 11, 2006). "Genetics of Breast and Ovarian Cancer (PDQ®) " Retrieved February 8, 2006, from <http://www.nci.nih.gov/cancertopics/pdq/genetics/breast-and-ovarian/HealthProfessional/page4>.
- National Cancer Institute. "Breast Cancer." Retrieved February 17, 2006, from <http://www.cancer.gov/cancertopics/types/breast>.
- Nelson, H. D., L. H. Huffman, et al. (2005). "Genetic risk assessment and BRCA mutation testing for breast and ovarian cancer susceptibility: systematic evidence review for the U.S. Preventive Services Task Force." Ann Intern Med 143(5): 362-79.
- Pharoah, P. D., A. Antoniou, et al. (2002). "Polygenic susceptibility to breast cancer and implications for prevention." Nat Genet 31(1): 33-6.
- Pinsky, L. E., J. B. Culver, et al. (2001). "Why should primary care physicians know about breast cancer genetics?" West J Med 175(3): 168-73.
- Stratton, J. F., P. Pharoah, et al. (1998). "A systematic review and meta-analysis of family history and risk of ovarian cancer." Br J Obstet Gynaecol 105(5): 493-9.

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