

American College of Obstetricians and Gynecologists (ACOG): Risk Assessment for Hereditary Breast and Ovarian Cancer Recommended

“Evaluating a patient’s risk for hereditary breast and ovarian cancer syndrome should be a routine part of obstetric and gynecologic practice.”¹

Mutations in *BRCA1* and *BRCA2* account for the vast majority of women with hereditary breast and ovarian cancer (HBOC). “In the general population, it is estimated that 1 in 300 to 1 in 800 individuals carry a *BRCA1* or *BRCA2* mutation,”² while “an estimated 1 in 40 Ashkenazi Jewish individuals carry one of three founder mutations.”^{3,4} Inheriting a *BRCA1* or *BRCA2* mutation, either from the maternal or the paternal lineage, increases a woman’s lifetime “risk of developing breast cancer to 65–74%”^{5,6} A *BRCA1* mutation increases the “risk for developing ovarian cancer to 39–46%”, while a *BRCA2* mutation increases this “risk to 12–20%”^{5,6} Evaluating a woman’s risk for carrying one of these mutations is an important first step in cancer prevention and early detection.

ACOG 2009 Practice Bulletin: Hereditary Breast and Ovarian Cancer Syndrome. *Obstet Gynecol.* 2009 Apr;113(4):957–66.

The ACOG recommends that evaluating a patient’s risk for hereditary breast and ovarian cancer syndrome be a routine part of obstetric and gynecologic practice. “Because tailored screening and prevention strategies can reduce morbidity and mortality from breast cancer and ovarian cancer, the identification of individuals at inherited risk is important.”¹

Similar to recommendations made by the Society of Gynecologic Oncology (SGO)⁷, the ACOG recommends genetic risk assessment for patients with a >20–25% chance of having an inherited predisposition to breast and ovarian cancer, also stating genetic risk assessment may still be helpful for patients with even a >5–10% chance of having this predisposition. These recommendations note that while testing an affected family member first is ideal, genetic testing may provide useful information for a patient without a personal history of cancer in the presence of a suggestive family history.

“Obstetricians and Gynecologists play an important role in the identification and management of women with hereditary breast and ovarian cancer syndrome.”¹

Risk assessment should be conducted by a health care provider with expertise in cancer genetics and includes gathering of family history information, risk assessment, education, and counseling.

A discussion should include:

- Possible test results: positive, negative, and uninformative, or variants of uncertain significance
- Options for medical management: surveillance, chemoprevention, risk-reducing surgeries
- Possible psychological and familial implications
- Cost of testing
- Insurance coverage for genetic testing
- Current legislation regarding genetic discrimination
- Privacy of genetic information

Genetic testing includes evaluation of the *BRCA1* and *BRCA2* genes for most patients, but in Ashkenazi Jewish individuals it often begins with testing for the three founder mutations. Once genetic test results are available, patients who test positive should be offered medical management options to reduce their risks of breast, fallopian tube, and ovarian cancer. ACOG provides additional guidance on surgical management of ovarian cancer risk, including discussion of risk-reducing salpingo-oophorectomy for *BRCA* positive women by the age of 40 years (or upon completion of child bearing) as well as information on the surgical technique for performing this procedure. Those patients who test negative for a *BRCA* mutation should be managed based on their family history of cancer.

1. ACOG 2009 Practice Bulletin: Hereditary Breast and Ovarian Cancer Syndrome. *Obstet Gynecol* 2009;113(4):957–66. 2. Whittemore AS, et al. Prevalence and contribution of *BRCA1* mutations in breast cancer and ovarian cancer: results from three U.S. population-based case control studies of ovarian cancer. *Am J Hum Genet* 1997;60:496–504. 3. Struwing JP, et al. The risk of cancer associated with specific mutations of *BRCA1* and *BRCA2* among Ashkenazi Jews. *N Engl J Med* 1997;336:1401–8. 4. Roa BB, et al. Ashkenazi Jewish population frequencies for common mutations in *BRCA1* and *BRCA2*. *Nat Genet* 1996;14:185–7. 5. Antoniou A, et al. Average risks of breast and ovarian cancer associated with *BRCA1* or *BRCA2* mutations detected in case series unselected for family history: a combined analysis of 22 studies [published erratum appears in *Am J Hum Genet* 2003;73:709]. *Am J Hum Genet* 2003;72:1117–30. 6. King MC, et al. Breast and ovarian cancer risks due to inherited mutations in *BRCA1* and *BRCA2*. New York Breast Cancer Study Group. *Science* 2003;302:643–6. 7. Society of Gynecologic Oncologists Education Committee statement on risk assessment for inherited gynecologic cancer predispositions. *Gynecol Oncol*;107:159–62.

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