

Clinical Summary: Risk Assessment, Genetic Counseling, and Genetic Testing for BRCA-Related Cancer

Population	Women with a personal or family history of breast, ovarian, tubal, or peritoneal cancer or who have an ancestry associated with <i>BRCA1/2</i> gene mutations	Women whose personal or family history or ancestry is not associated with potentially harmful <i>BRCA1/2</i> gene mutations
Recommendation	Assess with an appropriate brief familial risk assessment tool. Grade: B	Do not perform routine risk assessment, genetic counseling, or genetic testing. Grade: D

Risk Assessment	Patients with family or personal histories of breast, ovarian, tubal, or peritoneal cancer or ancestry associated with harmful <i>BRCA1/2</i> mutations should be assessed using a familial risk assessment tool. The USPSTF found adequate evidence that these tools are accurate in identifying women with increased likelihood of <i>BRCA1/2</i> mutations. Tools evaluated by the USPSTF include the Ontario Family History Assessment Tool, Manchester Scoring System, Referral Screening Tool, Pedigree Assessment Tool, 7-Question Family History Screening Tool, International Breast Cancer Intervention Study instrument (Tyrer-Cuzick), and brief versions of BRCAPRO. These tools should be used to guide referrals to genetic counseling.
Genetic Counseling	Genetic counseling about <i>BRCA1/2</i> mutation testing should be done by trained health professionals, including suitably trained primary care providers. The process of genetic counseling includes detailed kindred analysis and risk assessment for potentially harmful <i>BRCA1/2</i> mutations. It also includes identification of candidates for testing, patient education, discussion of the benefits and harms of genetic testing, interpretation of results after testing, and discussion of management options.
Genetic Testing	Tests for <i>BRCA1/2</i> mutations are highly sensitive and specific for known mutations. Testing for <i>BRCA1/2</i> mutations should be done when an individual has personal or family history that suggests an inherited cancer susceptibility, when an individual is willing to see a health professional who is suitably trained to provide genetic counseling and interpret test results, and when test results will aid in decision making.
Treatment and Interventions	In general, women with harmful <i>BRCA1/2</i> mutations are managed with a variety of interventions to lower future cancer risk. This includes intensive screening, risk-reducing medications, and risk-reducing mastectomy and salpingo-oophorectomy.
Relevant USPSTF Recommendations	The USPSTF recommends that clinicians offer to prescribe risk-reducing medications such as tamoxifen, raloxifene, or aromatase inhibitors to women at increased risk for breast cancer and at low risk for adverse medication effects. It recommends against the routine use of medications for risk reduction of primary breast cancer in women not at increased risk for breast cancer. The USPSTF recommends against screening for ovarian cancer in women. This recommendation does not apply to women with known genetic mutations that increase their risk for ovarian cancer (eg, <i>BRCA1/2</i> mutations). The USPSTF found insufficient evidence to assess the balance of benefits and harms of performing screening pelvic examinations in asymptomatic women for the early detection and treatment of a range of gynecologic conditions.

For a summary of the evidence systematically reviewed in making this recommendation, the full recommendation statement, and supporting documents, please go to <https://www.uspreventiveservicestaskforce.org>.