**Evidence-based Practice Guidelines Supporting Genetic Susceptibility Testing for Hereditary Breast and Ovarian Cancer Syndrome**

**United States Preventive Services Task Force (USPSTF) Recommendations (2019)1**

The USPSTF recommends that “primary care clinicians assess women with a personal or family history of breast, ovarian, tubal, or peritoneal cancer or who have an ancestry associated with *BRCA1/2* gene mutations with an

appropriate brief familial risk assessment tool.”

* Family history screening tools include
  + [Ontario Family History Assessment Tool](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab1)
  + [Manchester Scoring System](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab2)
  + [Referral Screening Tool](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab3)
  + [Pedigree Assessment Tool](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab4)
  + [7-Question Family History Screening Tool](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab5)
  + [International Breast Cancer Intervention Study instrument (Tyrer-Cuzick)](https://www.uspreventiveservicestaskforce.org/Page/Document/RecommendationStatementFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing1#tab6)
  + Brief versions of BRCAPRO
* “Women with a positive result on the risk assessment tool should receive genetic counseling and, if indicated after counseling, genetic testing.”
* The USPSTF recommendation includes the following personal or family health histories as examples of increased likelihood of having a *BRCA* mutation:
  + Breast cancer diagnosed before age 50
  + Bilateral breast cancer
  + Presence of both breast and ovarian cancer in the same person
  + Breast cancer in a male
  + Multiple cases of breast cancer in the family
  + One or more family members with two primary types of *BRCA*-related cancer
  + Ashkenazi (Eastern European) Jewish ancestry
  + Family member with a known harmful genetic mutation in the *BRCA1* or *BRCA2* gene

**Other recommendations**

* National Comprehensive Cancer Network (NCCN) (2019) Recommendations3
  + Referral for *BRCA* genetic counseling is recommended for individuals with a personal history of any of the following or a first- or second-degree relative (mother, father, brother, sister, child, grandparent, aunt, uncle, niece, or nephew) with any of the following:
    - Family member with a known pathogenic or likely pathogenic *BRCA* mutation
    - Ovarian carcinoma
    - Male breast cancer
    - Pancreatic cancer
    - Metastatic prostate cancer
    - Female breast cancer and one or more of the following
      * Diagnosed ≤ age 45
      * Diagnosed age 46-50 with:
        + Another breast cancer primary at any age
        + ≥ 1 close blood relative with breast cancer or high grade (Gleason score ≥7) prostate cancer at any age
        + An unknown or limited family history
      * Diagnosed ≤ age 60 with triple negative breast cancer
      * Diagnosed at any age with
        + ≥ 1 close blood relative with

breast cancer diagnosed ≤ age 50

ovarian carcinoma

male breast cancer

metastatic prostate cancer

pancreatic cancer

* + - * + ≥ 2 close blood relatives with breast cancer at any age
        + ≥ 2 additional breast cancer primaries at any age
        + Ashkenazi or Eastern European Jewish ancestry
    - High grade prostate cancer (Gleason score ≥7) at any age and one or more of the following
      * ≥ 1 close blood relatives with
        + Ovarian carcinoma at any age
        + Pancreatic cancer at any age
        + Metastatic prostate cancer at any age
        + Breast cancer ≤50 years
      * ≥ 2 close blood relatives with
        + Breast cancer at any age
        + Prostate cancer (any grade) at any age
      * Ashkenazi (Eastern European) Jewish ancestry
    - BRCA1/2 pathogenic or likely pathogenic mutation detected by tumor profiling in the absence of germline mutation analysis
* American College of Medical Genetics (ACMG) and National Society of Genetic Counselors (NSGC) Recommendations (2014)4
  + Referral for genetic counseling for *BRCA* is recommended for individuals with a personal or family heath history of
    - Breast cancer diagnosed ≤ age 50
    - Triple-negative breast cancer diagnosed ≤ age 60
    - Two or more primary breast cancers
    - Ovarian, fallopian tube, or primary peritoneal cancer
    - Male breast cancer
    - Ashkenazi Jewish or Eastern European ancestry and breast or pancreatic cancer at any age
    - Breast, ovarian, or pancreatic cancer and two or more cases of breast, ovarian, pancreatic, or aggressive prostate cancer in close blood relatives Aggressive prostate cancer and two or more cases of breast, ovarian, or pancreatic cancer in close blood relatives
    - Aggressive prostate cancer and two or more cases of breast, ovarian, or pancreatic cancer in close blood relatives