

Hereditary Breast & Ovarian Cancer and BRCA1/2 Testing

Overview

Hereditary breast and ovarian cancer syndromes are primarily associated with pathogenic variants in the BRCA1 and BRCA2 genes. Women carrying these mutations have up to a 65–80% lifetime risk of breast cancer and 20–40% risk of ovarian cancer. Men with BRCA mutations are also at elevated risk for prostate, pancreatic, and male breast cancers.

Early identification of BRCA mutation carriers allows for targeted prevention, surveillance, and treatment strategies that can significantly reduce morbidity and mortality.

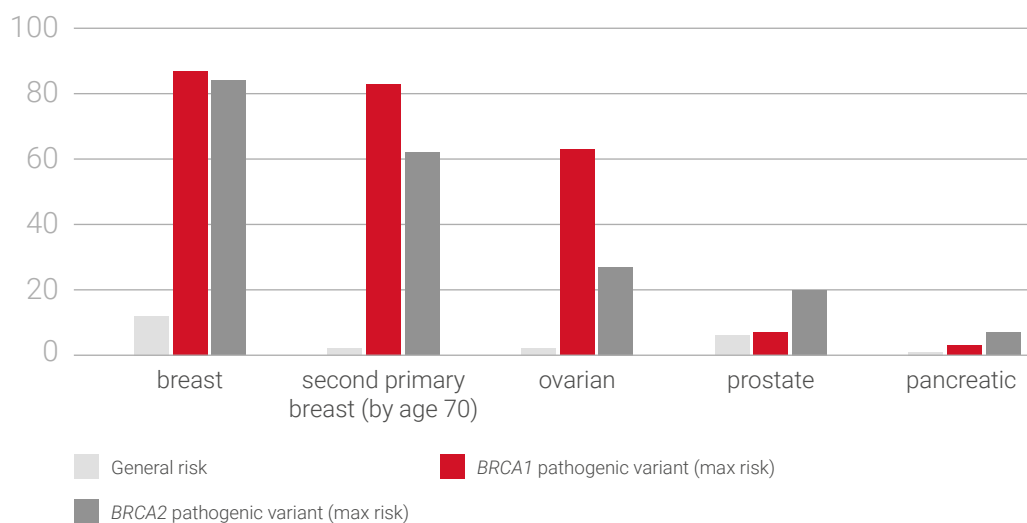
Who Should Be Tested?

Testing should be considered for individuals with:

- Personal history of breast cancer diagnosed before age 50.
- Triple-negative breast cancer diagnosed before age 60.
- Personal or family history of ovarian, fallopian tube, or peritoneal cancer.
- Male breast cancer at any age.
- Multiple family members with breast, ovarian, prostate, or pancreatic cancers.
- Known BRCA1/2 mutation in the family.

Testing is also appropriate for unaffected individuals with a strong family history suggestive of hereditary cancer predisposition.

BRCA1/2 increases the lifetime risk of developing cancer



Test Methodology

Farabi Medical Laboratory uses Next-Generation Sequencing (NGS) to analyze the full coding regions and intron–exon boundaries of BRCA1 & BRCA2 genes. Large genomic rearrangements (deletions / duplications) are also detected using MLPA or CNV analysis.

Clinical Management of Mutation Carriers

Positive test results should be followed by genetic counseling and personalized risk management, which may include:

- Enhanced surveillance: annual MRI and mammography beginning at 25–30 years.
- Risk-reducing surgery: prophylactic mastectomy and/or salpingo-oophorectomy.
- Chemoprevention: selective estrogen receptor modulators (SERMs).
- Targeted therapy: PARP inhibitors (olaparib, talazoparib) for BRCA-associated cancers.

Implications for Family Members

Because BRCA mutations are autosomal dominant, each first-degree relative has a 50% chance of carrying the variant. Cascade testing of relatives allows early risk identification and prevention.

Interpreting Test Results

Result Type	Meaning	Next Steps
Pathogenic / Likely Pathogenic Variant	Increased cancer risk confirmed	Genetic counseling, surveillance, preventive measures
Variant of Uncertain Significance (VUS)	Unclear clinical impact	Periodic re-evaluation; no management change until clarified
Negative Result	No known pathogenic variant detected	Standard population risk management

Why Choose Farabi Medical Laboratory

- Advanced NGS-based BRCA testing with confirmatory CNV analysis
- Accredited molecular laboratory based in Erbil, Iraq
- Comprehensive genetic counseling support
- Fast turnaround time and local clinician collaboration