Breast Cancer Management Panel

BioReference LABORATORIES | genpath | GeneDx
OPKO Health Companies

Hereditary Breast Cancer

Genetic testing with the Breast Cancer Management Panel may be appropriate if your personal and/or family history is suggestive of a hereditary predisposition to cancer. **This includes:**

- Breast cancer diagnosed under the age of 50 or triple negative breast cancer diagnosed under the age of 60
- Multiple cancers in one person, either of same origin (such as two separate breast cancers) or of different origins (such as breast and ovarian cancer)
- Diagnosis of ovarian cancer*, pancreatic cancer*, metastatic prostate cancer*, or male breast cancer at any age
- Multiple relatives diagnosed with the same or related cancers (including breast, ovarian, pancreatic, and/or metastatic/aggressive prostate cancer) on the same side of the family and spanning multiple generations
- Ashkenazi Jewish ancestry
- A variant in *BRCA1* or *BRCA2* identified on tumor sequencing

Your healthcare provider will determine if genetic testing is medically necessary for you.

*If this is the primary indication for testing, a more comprehensive panel specifically geared at this diagnosis is available

Genes Included on the Breast Cancer Management Panel are Listed in the Table Below

High-Risk Genes — Well-studied • Greater than 4-fold risk of developing one or more cancers • Can cause a moderate risk for other cancers • National or expert opinion guidelines for screening and prevention are established

Moderate-Risk Genes — Well-studied • Approximately 2- to 4-fold risk of developing one or more cancers • May increase risk for other cancers • Limited guidelines for screening and prevention

Newer Genes — Not as well-studied • Precise lifetime risks and tumor spectrum not yet determined • Guidelines for screening and prevention are limited or not available

	Gene	Lifetime Cancer and/or Tumor Risks*
High-Risk Genes	*BRCA1*	Female breast (43-87%), Ovarian, including primary peritoneal and fallopian tube (39-63%), Prostate, Male breast (1.2%), Pancreatic (1-3%)
	BRCA2	Female breast (38-84%), Ovarian, including primary peritoneal and fallopian tube (16.5-27%), Prostate (up to 20%), Pancreatic (2-7%), Male breast (up to 8.9%), Melanoma
	CDH1	Gastric-diffuse, Female breast-lobular (42-55%), Colorectal
	PALB2	Female breast (33-53%), Male breast, Pancreatic (2-4%), Ovarian, Prostate
	PTEN	Female breast (25-85%), Thyroid (3-38%), Endometrial (5-28%), Colorectal, Renal, Melanoma, Gastrointestinal polyps, Lhermitte-Duclos disease
	STK11	Female breast (up to 54%), Colorectal (39%), Pancreatic (11-36%), Gastric (29%), Ovarian tumors (21%), Lung (7-17%), Small bowel (13%), Cervical (10%), Testicular tumors (9%), Endometrial (9%), Gastrointestinal polyps
	TP53	Female breast (85%), Soft tissue sarcoma, Osteosarcoma, Brain, Hematologic malignancies-Acute leukemias among others, Adrenocortical carcinoma, among others. Overall risk for cancer: approaches 100% in females, 73% in males
Moderate-Risk Genes	*ATM*	Female breast (27-33%), Colorectal, Ovarian, Pancreatic, Prostate
	CHEK2	Female breast, Male breast, Colorectal, Gastric, Prostate, Renal, Thyroid
Newer Genes	*BARD1*	Female breast

*Most commonly associated cancer/tumors listed; lifetime risks provided when available. Risks relate to carriers of a single pathogenic variant.

Possible Outcomes of Genetic Testing

+ Positive

- Pathogenic or likely pathogenic variant identified
- Medical management recommendations may be available
- Family member testing may be recommended

− Negative

- No significant genetic changes identified
- Medical management based on personal and/or family history

? Variant of Uncertain Significance (VUS)

- A genetic change identified, but its association with disease is unclear
- Medical management based on personal and/or family history

Medical Management Based on Genetic Test Results

Clinical guidelines may be available which provide options and recommendations for patients who have a positive (pathogenic variant or likely pathogenic) test result indicating an increased risk for cancer and/or tumors. Guidelines and recommendations for early detection and/or risk reduction are specific to the gene in which the pathogenic variant was found.

Recommendations May Include:

- Breast awareness, including breast self-examination for both men and women
- Clinical exams, such as skin and/or breast exams
- Imaging exams, such as mammogram, MRI, CT and/or ultrasound
- Screening procedures, such as colonoscopy
- Risk-reducing medications and/or surgeries

In some cases, guidelines for screening and prevention are limited or not available for a positive result. Once your test results are available, a discussion with your healthcare provider is recommended to determine the most appropriate medical management options for you and your family.

Resources

General
American Cancer Society
www.cancer.org

GeneDx
www.genedx.com/oncology

National Cancer Institute
www.cancer.gov

Breast Cancer
Bright Pink
www.brightpink.org

Facing Our Risk of Cancer Empowered (FORCE)
www.facingourrisk.org

Find a Genetic Counselor
Canadian Association of Genetic Counsellors
www.cagc-accg.ca

National Society of Genetic Counselors
www.nsgc.org