**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**

<table>
<thead>
<tr>
<th>High-Risk Genes</th>
<th>Well-studied</th>
<th>Greater than 4-fold risk of developing one or more cancers</th>
<th>Can cause a moderate risk for other cancers</th>
<th>National or expert opinion guidelines for screening and prevention are established</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>BRCA1</strong></td>
<td>Female breast (55-87%), Ovarian (39-59%), Prostate, Male breast, Pancreatic, Fallopian tube, Primary peritoneal, Endometrial-serous</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>BRCA2</strong></td>
<td>Female breast (33-84%), Prostate (up to 34%), Ovarian (11-27%), Pancreatic (up to 7%), Male breast (up to 7%), Melanoma, Fallopian tube, Primary peritoneal, Endometrial-serous</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>CDH1</strong></td>
<td>Gastric-diffuse, Female breast-lobular (39-55%), Colorectal</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>PALB2</strong></td>
<td>Female breast (up to 58%), Male breast, Pancreatic, Ovarian, Prostate</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>PTEN</strong></td>
<td>Female breast (25-85%), Thyroid (3-38%), Endometrial (5-28%), Colorectal, Renal, Melanoma, Gastrointestinal polyps, Lhermitte-Duclos disease</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>TP53</strong></td>
<td>Female breast (85%), Soft tissue sarcoma, Osteosarcoma, Brain, Hematologic malignancies-Acute leukemias among others, Adrenocortical carcinoma, among others. Overall risk for cancer: up to 95% in females, 88% in males</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Moderate-Risk Genes</th>
<th>Well-studied</th>
<th>Approximately 2- to 4-fold risk of developing one or more cancers</th>
<th>May increase risk for other cancers</th>
<th>Limited guidelines for screening and prevention</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>ATM</strong></td>
<td>Female breast (27-33%), Colorectal, Ovarian, Pancreatic, Prostate</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>CHEK2</strong></td>
<td>Female breast, Male breast, Colorectal, Gastric, Prostate, Renal, Thyroid</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Newer Genes</th>
<th>Not as well-studied</th>
<th>Precise lifetime risks and tumor spectrum not yet determined</th>
<th>Guidelines for screening and prevention are limited or not available</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>NBN</strong></td>
<td>Female breast, Non-Hodgkin lymphoma, Prostate</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

*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](http://www.cancer.org)
- GeneDx
  [www.genedx.com/oncology](http://www.genedx.com/oncology)
- National Cancer Institute
  [www.cancer.gov](http://www.cancer.gov)

**Breast Cancer**
- Bright Pink
  [www.brightpink.org](http://www.brightpink.org)
- Facing Our Risk of Cancer Empowered (FORCE)
  [www.facingourrisk.org](http://www.facingourrisk.org)

**Find a Genetic Counselor**
- Canadian Association of Genetic Counsellors
  [www.caagc.ca](http://www.caagc.ca)
- National Society of Genetic Counselors
  [www.nsgc.org](http://www.nsgc.org)