Understanding Your Positive BRCA1 Genetic Test Result
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 THINGS TO KNOW

1. **BRCA1 mutation**
   Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the **BRCA1** gene.

2. **Hereditary breast and ovarian cancer (HBOC)**
   People with **BRCA1** mutations have hereditary breast and ovarian cancer (HBOC).

3. **Cancer risks**
   You have an increased chance to develop female or male breast cancer, ovarian cancer, pancreatic cancer, prostate cancer, and possibly other types of cancer.

4. **What you can do**
   There are risk management options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your doctor, and decide on a plan that best manages cancer risks.

5. **Family**
   Family members may also be at risk – they can be tested for the **BRCA1** mutation that was identified in you.

**BRCA1 MUTATION LIFETIME CANCER RISKS (%)**

<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>General Population</th>
<th>BRCA1 Mutation Carrier</th>
</tr>
</thead>
<tbody>
<tr>
<td>Female Breast Cancer</td>
<td>12</td>
<td>increased</td>
</tr>
<tr>
<td>Ovarian Cancer</td>
<td>2</td>
<td>increased</td>
</tr>
<tr>
<td>Male Breast Cancer</td>
<td>0.1</td>
<td>increased</td>
</tr>
<tr>
<td>Prostate Cancer</td>
<td>14</td>
<td>increased</td>
</tr>
<tr>
<td>Pancreatic Cancer</td>
<td>1.5</td>
<td>increased</td>
</tr>
</tbody>
</table>

**BRCA1 MUTATIONS IN THE FAMILY**

There is a 50/50 random chance to pass on a genetic mutation in **BRCA1** to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations.

*The above cancer risks represent the typical range for individuals with a mutation in this gene. If available, cancer risks specific to the mutation found in you will be provided in your results report.*
Understanding Your Positive BRCA1 Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

RESULT | MUTATION | Your testing shows that you have a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or a variant that is likely pathogenic in the BRCA1 gene. Both of these results should be considered positive.

GENE | BRCA1 | Everyone has two copies of the BRCA1 gene, which we randomly inherit from each of our parents. Mutations in one copy of the BRCA1 gene can increase the chance for you to develop certain types of cancer in your lifetime.

CONDITION | HBOC | People with BRCA1 mutations have hereditary breast ovarian cancer (HBOC).

CANCER RISKS | INCREASED | You have an increased chance to develop female or male breast cancer, ovarian, fallopian tube, or primary peritoneal cancer, pancreatic cancer, prostate cancer, and possibly other types of cancer.

MANAGEMENT OPTIONS | FOR WOMEN | Options for early detection and prevention for women may include: breast exam, mammogram, breast MRI, transvaginal ultrasound, a blood test called CA-125, preventive medications, and options for preventive surgery. Talk to your doctor about what options may be right for you.

MANAGEMENT OPTIONS | FOR MEN | Options for screening and early detection for men may include: breast exam, mammogram, and increased prostate screening. Talk to your doctor about what options may be right for you.

RISK MANAGEMENT | VARIES | Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than the general population and is often more frequently performed. It is important to discuss these options with your doctor.

FAMILY MEMBERS | 50/50 CHANCE | Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the BRCA1 mutation that you carry, and other family members (like your aunts, uncles, cousins) may also inherit it. Your relatives can be tested for this same mutation. Depending on the family history, those who DO NOT have it may not have an increased chance (above the general population) to develop cancer.

NEXT STEPS | DISCUSS | It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

REACH OUT | RESOURCES | • Ambry’s hereditary cancer site for families patients.ambrygen.com/cancer
• Bright Pink brightpink.org
• FORCE facingourrisk.org
• Sharsheeret sharsheeret.org
• Susan G. Komen Foundation komen.org
• Genetic Information Nondiscrimination Act (GINA) ginahelp.org
• National Society of Genetic Counselors nsgc.org
• Canadian Association of Genetic Counselors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your BRCA1 result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.

WATCH THE VIDEO
Scan or visit our website to watch a short video with additional information to help you understand your results. ambrygen.com/cancer/uyr/brca1