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

4 THINGS TO KNOW

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

2. **Cancer risks**
   - You have an increased chance to develop female breast cancer and possibly other cancers such as ovarian cancer.

3. **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.

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

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**NBN MUTATIONS IN THE FAMILY**

There is a 50/50 random chance to pass on a mutation in NBN to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.
## Understanding Your Positive NBN Genetic Test Result

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

<table>
<thead>
<tr>
<th>RESULT</th>
<th>MUTATION</th>
<th>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 NBN gene. Both of these results should be considered positive.</th>
</tr>
</thead>
<tbody>
<tr>
<td>GENE</td>
<td>NBN</td>
<td>Everyone has two copies of the NBN gene, which we randomly inherit from each of our parents. Mutations in one copy of the NBN gene can increase the chance for you to develop certain types of cancer in your lifetime.</td>
</tr>
<tr>
<td>CANCER RISKS</td>
<td>INCREASED</td>
<td>You have an increased lifetime chance to develop female breast cancer and possibly other cancers such as ovarian cancer, medulloblastoma (a cancer of the brain) and/or other tumors of the central nervous system. Men have an increased chance to develop prostate cancer.</td>
</tr>
<tr>
<td>OTHER MEDICAL CONCERNS</td>
<td>MAY BE PRESENT</td>
<td>Individuals with NBN mutations may have an increased risk (25%) to have a child with Nijmegen breakage syndrome, but only if their partner also carries a mutation in the NBN gene. Nijmegen breakage syndrome is a rare condition that may cause limited growth in childhood, immunodeficiency, and other medical concerns including increased cancer risks.</td>
</tr>
<tr>
<td>MANAGEMENT OPTIONS</td>
<td>FOR WOMEN</td>
<td>Options for early detection and prevention for women depend on your family history of cancer and may include: breast exam, mammogram, breast MRI, and options for preventive surgery. Talk to your doctor about what options may be right for you.</td>
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<tr>
<td>RISK MANAGEMENT</td>
<td>VARIES</td>
<td>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. Individuals with NBN mutations should also avoid sun exposure and protect their skin when outside.</td>
</tr>
<tr>
<td>FAMILY MEMBERS</td>
<td>50/50 CHANCE</td>
<td>Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the NBN 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.</td>
</tr>
<tr>
<td>NEXT STEPS</td>
<td>DISCUSS</td>
<td>It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.</td>
</tr>
</tbody>
</table>
| REACH OUT | RESOURCES | • Ambry’s hereditary cancer site for families [patients.ambrygen.com/cancer](http://patients.ambrygen.com/cancer)  
• American Cancer Society [cancer.org](http://cancer.org)  
• FORCE [facingourrisk.org](http://facingourrisk.org)  
• Genetic Information Nondiscrimination Act (GINA) [ginahelp.org](http://ginahelp.org)  
• National Society of Genetic Counselors [nsgc.org](http://nsgc.org)  
• Canadian Association of Genetic Counsellors [cagc-accg.ca](http://cagc-accg.ca)  

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your NBN 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.