Understanding Your Positive BRIP1 Genetic Test Result
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

4 THINGS TO KNOW

1. BRIP1 mutation
   Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the BRIP1 gene.

2. Cancer risks
   You have an increased chance to develop ovarian cancer, and possibly other cancers such as female breast cancer. Cancer risk estimates for male BRIP1 mutation carriers are not currently available.

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

4. Family
   Family members may also be at risk – they can be tested for the BRIP1 mutation that was identified in you.

BRIP1 MUTATIONS IN THE FAMILY

There is a 50/50 random chance to pass on a mutation in BRIP1 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 \textit{BRIP1} Genetic Test Result

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

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- 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 \textit{BRIP1} gene. Both of these results should be considered positive.

- Everyone has two copies of the \textit{BRIP1} gene, which we randomly inherit from each of our parents. Mutations in one copy of the \textit{BRIP1} gene can increase the chance for you to develop certain types of cancer in your lifetime.

- You have an increased chance to develop ovarian cancer (up to 9%, compared to 2% in the average woman), as well as an increased chance to develop female breast cancer. Cancer risk estimates for male \textit{BRIP1} mutation carriers are not currently available.

- Individuals with \textit{BRIP1} mutations may have an increased risk (25%) to have a child with Fanconi anemia, but only if their partner also carries a mutation in the \textit{BRIP1} gene. Fanconi anemia is a rare condition that can cause specific physical characteristics, bone marrow failure, and an increased risk of certain cancers.

- Options for screening and early detection of cancer for women may include: breast exam, mammogram, breast MRI, and options for preventive surgeries. Talk with your doctor about what options may be right for you.

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

- Your close relatives (like your parents, brothers, sisters, and children) have a 50/50 random chance of inheriting the \textit{BRIP1} 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.

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

**RESOURCES**

- Ambry’s hereditary cancer site for families: patients.ambrygen.com/cancer
- American Cancer Society: cancer.org
- FORCE: facingourrisk.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 \textit{BRIP1} 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.