Understanding Your Positive BAP1 Genetic Test Result
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

4 THINGS TO KNOW

1. **BAP1 mutation**
   
   Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the BAP1 gene.

2. **Cancer risks**
   
   You have an increased chance to develop mesothelioma, kidney (renal) cancer, melanoma (skin cancer), uveal (eye) melanoma, and other tumor types.

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 BAP1 mutation that was found in you.

**BAP1 MUTATIONS IN THE FAMILY**

There is a 50/50 random chance to pass on a BAP1 mutation 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 BAP1 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 variant that is likely pathogenic in the **BAP1** gene. Both of these results should be considered positive.

- Everyone has two copies of the **BAP1** gene, which we randomly inherit from each of our parents. Mutations in one copy of the **BAP1** gene can increase the chance for you to develop certain types of cancer in your lifetime.

- You have an increased chance to develop mesothelioma (cancer of the protective lining that covers the lungs, stomach, and other organs), which is often caused by asbestos exposure. You also have an increased chance to develop kidney cancer, melanoma (skin cancer), uveal (eye) melanoma, and other tumor types.

- Options for screening and early detection may include screening of the kidneys, such as ultrasounds or MRI, skin examinations by a dermatologist, and eye evaluations by an ophthalmologist. Talk to your doctor about which 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 more frequently done. It is important to discuss these options with your doctor.

- Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the **BAP1** 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 lifetime 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**
  - American Cancer Society: cancer.org
  - Genetic Information Nondiscrimination Act (GINA): ginahelp.org
  - National Society of Genetic Counselors: nsgc.org
  - Canadian Association of Genetic Counsellors: cagc-accg.ca
  - Research study: Do **BAP1** mutation carriers have increased sensitivity to radiation? Contact: Dr. Friedman feitan@post.tau.ac.il

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your **BAP1** 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.