**BAP1**

The BAP1 gene is a tumor suppressor gene. Tumor suppressor genes slow down cell division, repair DNA mistakes, or tell cells when to die. When they don't work properly, cells can grow out of control, which can lead to cancer. The primary roles of BAP1 are to assist in the repair of damaged DNA and to regulate cell growth and division.

Like most genes, each person has two copies of the BAP1 gene: one inherited from each parent. A mutation in a single BAP1 gene inherited from either parent is known to increase risks of melanoma of the skin (cutaneous melanoma) and eye (uveal melanoma), as well as kidney and lung cancer (specifically a type called mesothelioma) and non-cancerous skin growths called melanocytic BAP1-mutated atypical intradermal tumors (MBAITs).

Certain factors can greatly increase risk of melanoma, including an individual's geographic region, ethnicity and sun exposure. For example, melanoma is 20 times more common in Caucasians than it is in African Americans.\(^1\) The risk of kidney cancer also varies depending on whether a person has a history of smoking cigarettes or exposure to certain substances and chemicals.\(^2\) The risk of lung (mesothelioma) cancer is impacted by exposure to asbestos and other harmful substances.\(^3\)

**How common are mutations in the BAP1 gene?**

Mutations in the BAP1 gene are rare—the exact frequency is not yet known. Studies to establish the frequency of BAP1 mutations are ongoing.

**How mutations in this gene impact risk**

**Women**

If a woman has a mutation in the BAP1 gene, her chances of developing melanoma, kidney, and lung cancer (specifically a type called mesothelioma), are greater than that of the average US woman. This does not mean that she has a diagnosis of cancer or that she will definitely develop cancer in her lifetime.

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### Cancer by age 95

<table>
<thead>
<tr>
<th></th>
<th>Average US woman</th>
<th>With BAP1 mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Melanoma</strong></td>
<td>1.6%</td>
<td>Elevated(^5,6)</td>
</tr>
<tr>
<td><strong>Kidney</strong></td>
<td>1.2%</td>
<td>Elevated(^5,7)</td>
</tr>
<tr>
<td><strong>Lung (mesothelioma)</strong></td>
<td>&lt;1%</td>
<td>Elevated(^5,6)</td>
</tr>
</tbody>
</table>

*Elevated: Risk is increased, but further research may clarify the exact risk figure.*

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### Men

If a man has a mutation in the BAP1 gene, his chances of developing melanoma, kidney, and lung cancer (specifically a type called mesothelioma), are greater than that of the average US man. This does not mean that he has a diagnosis of cancer or that he will definitely develop cancer in his lifetime.

<table>
<thead>
<tr>
<th></th>
<th>Average US man</th>
<th>With BAP1 mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Melanoma</strong></td>
<td>2.6%</td>
<td>Elevated(^5,6)</td>
</tr>
<tr>
<td><strong>Kidney</strong></td>
<td>2%</td>
<td>Elevated(^5,7)</td>
</tr>
<tr>
<td><strong>Lung (mesothelioma)</strong></td>
<td>&lt;1%</td>
<td>Elevated(^5,6)</td>
</tr>
</tbody>
</table>

*Elevated: Risk is increased, but further research may clarify the exact risk figure.*

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### Screening guidelines

Below is a summary of screening guidelines from the American Cancer Society (ACS). Because there are no published screening guidelines specific to individuals with BAP1 mutations, these guidelines are for individuals who have the same risk of melanoma as the average US individual. If you have a mutation in this gene, your healthcare provider may use these ACS Guidelines to help create a customized screening plan for you. They might also make additional recommendations to reduce the risk of melanoma.

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Women and Men

Melanoma

- Your healthcare provider may discuss skin exams and eye exams to screen for melanoma.
- To reduce the chance of developing melanoma, the American Cancer Society recommends limiting exposure to UV light by avoiding excess sun exposure, wearing a hat, sunglasses and long protective clothing, applying sunscreen with SPF of 30 or higher and avoiding tanning beds and sun lamps.
- Any new, unusual, or changing moles should be reported to your provider or dermatologist.

Kidney cancer

- Currently, there are no kidney cancer screening guidelines specific to \textit{BAP1} mutation carriers. Your provider may discuss screening or referral to a specialist.

Lung cancer (mesothelioma)

- Currently, there are no lung cancer screening guidelines specific to \textit{BAP1} mutation carriers. Your provider may discuss screening or referral to a specialist.

Useful resources

American Melanoma Foundation
An organization supporting melanoma research, and providing advocacy and public awareness of melanoma.

\url{www.melanomafoundation.org}

Kintalk
An educational and family communication site for individuals and their families with hereditary cancer conditions.

\url{www.kintalk.org}

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