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Clinician Management Resource for *BAP1*

This overview of clinical management guidelines is based on this patient's positive test result for *BAP1* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those published in GeneReviews[1]. Please consult the referenced website link for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries, and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decision but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider and may change.

SURVEILLANCE CONSIDERATIONS[1,^]	AGE TO START	FREQUENCY
***BAP1*-inactivated melanocytic tumor, cutaneous melanoma, and/or basal cell carcinoma**		
Full-body skin exam by dermatologist specializing in melanoma	Beginning at age ~18 years	Annually
Consider whole-body imaging for patients with a large number of lesions. Whole-body imaging is recommended for stable asymptomatic *BAP1*-inactivated melanocytic tumor lesions.	Beginning at age ~18 years	Annually
Uveal melanoma		
Dilated eye exam and baseline dilated fundus imaging, preferably by an ophthalmologist trained in diagnosis and management of uveal melanoma (ocular oncologist)	Beginning at age ~11 years	Annually
Refer any suspected lesion to ophthalmologist specializing in management of uveal melanoma (ocular oncologist) for proper diagnosis and management. If diagnosed, manage uveal melanoma as a more aggressive tumor due to the increased aggressiveness of *BAP1*-related uveal melanoma.	Beginning at age ~11 years	Individualized
Malignant mesothelioma		
Clinical evaluation for signs/symptoms of pleurisy, peritonitis, ascites, and/or pleural effusion, such as: chest pain, cough, fever, shortness of breath, dysphagia, hoarseness, weight loss, fever, upper body and face edema, abdominal pain, nausea, vomiting, and/or constipation	Beginning at age 30 years	Individualized
Asymptomatic imaging surveillance with ultrasound (renal/abdominal and chest) or MRI (abdominal and chest with diffusion-weighted sequences)	Beginning at age 30 years*	Individualized*
Renal cell carcinoma		
Clinical abdominal exam with investigation of any suspected symptoms, such as abdominal pain and/or hematuria	Beginning at age 30 years	Individualized
Asymptomatic imaging surveillance using ultrasound (renal/abdominal and chest) and MRI (abdominal and chest with diffusion-weighted sequences)	Beginning at age 30 years**	Every 2 years**
Counseling		
Counsel patients to avoid the following agents/circumstances: arc welding, asbestos, smoking, unnecessary and prolonged sun exposure, routine chest x-ray and CT examinations	Individualized	Individualized
Genetic counseling by genetics professionals is recommended to inform affected persons and their families regarding the nature, mode of inheritance, and implications of *BAP1*-TPDS to facilitate medical and personal decision making.	Individualized	Individualized

^ To establish the extent of disease and needs in an individual diagnosed with *BAP1* tumor predisposition syndrome, the evaluations summarized in the table above are recommended in a multidisciplinary team approach.

* Combined with renal cell carcinoma evaluation. If abdominal MRI is to be performed as recommended for renal cell carcinoma, consider evaluation of peritoneum and pleura as well. Some physicians recommend spiral chest CT for asymptomatic persons with a history of exposure to asbestos, while others do not, given the possible increased risk of cancer from radiation exposure. Avoid routine surveillance with chest x-ray or CT exam.

** Combined with malignant mesothelioma exam. Follow-up MRI and ultrasound should be performed in alternating years.

1. Pilarski R, et al. 2016 Oct 13 [Updated 2022 Mar 24]. In: GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. https://www.ncbi.nlm.nih.gov/books/NBK390611/

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Understanding Your Positive *BAP1* Genetic Test Result

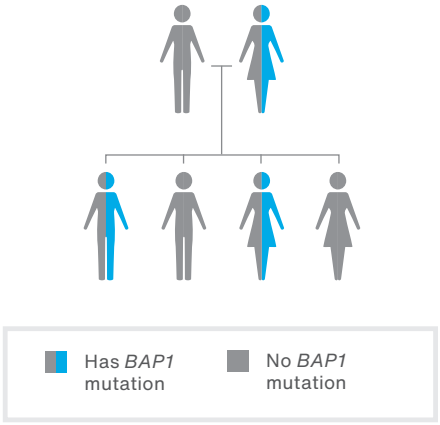
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

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 and other medical concerns	You have an increased chance to develop benign skin tumors (also called melanocytic tumors), uveal (eye) melanoma, mesothelioma (cancer of the protective lining that covers the lungs, stomach, and other organs), melanoma (skin cancer), and other tumor types.
3	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
4	Family	Family members may also be at risk – they can be tested for the *BAP1* mutation that was found in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

BAP1 Mutations in the Family

There is a 50/50 random chance to pass on a *BAP1* mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth.



Reach Out	RESOURCES	• American Cancer Society cancer.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.

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