Ambry Genetics®
A TEMPUS COMPANY

Clinician Management Resource for *BAP1*

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic *BAP1* variant. 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 a dermatologist specializing in melanoma, including exam of nails to evaluate for subungual tumors.	Beginning at age ~18 years	Annually
Consider total body photography for patients with a large number of lesions.	Beginning at age ~18 years	Individualized
Uveal melanoma		
Dilated eye exam and baseline dilated fundus imaging.	Beginning at age ~11-16 years, or at puberty	Annually
Referral to ophthalmologist specializing in management of uveal melanoma for any pigmented lesions.	Individualized	Individualized
Malignant mesothelioma*		
Clinical evaluation for manifestations 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	Annually
Abdominal MRI combined with renal cell carcinoma screening.** Consider chest and pelvic MRI as part of a clinical trial.	Beginning at age 30 years	Individualized
Renal cell carcinoma		
Clinical evaluation for manifestations of renal cell carcinoma, such as flank pain and/or hematuria.	Beginning at age 30 years	Annually
Abdominal ultrasound/MRI with diffusion-weighted sequences. Alternate ultrasound every two years with MRI every two years so that imaging is done annually.**	Beginning at age 30 years	Annually
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 a clinical geneticist, certified genetic counselor, certified genetic nurse, or genetics advanced practice provider to obtain a pedigree and inform affected individuals and their families regarding the nature, mode of inheritance, and implications of *BAP1* tumor predisposition syndrome to facilitate medical and personal decision making.	At diagnosis	Individualized

^ The evaluations summarized in the table above are also recommended to establish the extent of disease and needs in an individual diagnosed with *BAP1* tumor predisposition syndrome, if not performed as part of the evaluation that led to the diagnosis.

* No consensus on screening modalities for malignant mesothelioma exist.

** Abdominal MRI for evaluation of malignant mesothelioma should be combined with renal cell carcinoma evaluation. Avoid routine surveillance with chest radiograh or CT.

1. Pilarski R, et al. 2016 Oct 13 [Updated 2024 Dec 5]. In: GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. https://www.ncbi.nlm.nih.gov/books/NBK390611/

Understanding Your Positive *BAP1* Genetic Test Result

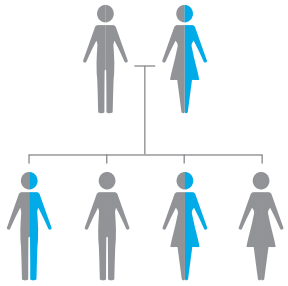
INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

4 Things to know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant 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 pathogenic or likely pathogenic *BAP1* variant 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 in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *BAP1* variant to each of your children.



Has a pathogenic or likely pathogenic *BAP1* variant

No pathogenic or likely pathogenic *BAP1* variant

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.

1 Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

MKT-ONCO-FLYR-10064-EN v6 11.19.25