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Clinician Management Resource for Birt-Hogg-Dubé syndrome

This overview of clinical management guidelines is based on this patient's positive test result for *FLCN* 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
Cutaneous manifestations		
Detailed skin exam by dermatologist to screen for possible risk of melanoma.	At diagnosis	Every 6-12 months
Lung cysts and/or pneumothorax		
High-resolution CT or CT of the chest for visualization of pulmonary cysts. Individuals with signs/symptoms of pneumothorax should immediately undergo chest x-ray and CT of the chest.	At diagnosis	Individualized* or for those with 1) suspected/treated pneumothorax; or 2) prior to anesthesia or long-distance flight
Renal tumors		
Abdominal/pelvic MRI or abdominal/pelvic CT with contrast if MRI is not an option.	At diagnosis, or beginning at age 20 years**	Annually, for patients with suspicious lesions. In patients with no personal or family history of renal tumors, screen every 2 years after 2-3 consecutive normal MRIs.
Parotid tumors		
Review signs/symptoms of parotid tumors	Individualized	Consider Annually
Thyroid cancer		
Thyroid ultrasound	Individualized	Consider annually
Colorectal cancer (CRC)		
Colonoscopy	Age 40 years or 10 years before youngest CRC diagnosis in family, whichever is earlier	Individualized
Counseling		
Counsel patients to avoid the following agents/circumstances: cigarette smoking, high ambient pressures, radiation exposure	Individualized	Individualized
Consultations with a genetic counselor and/or a clinical geneticist.	At diagnosis	Individualized

^ Includes recommended evaluations to establish the extent of disease in patients diagnosed with Birt-Hogg-Dubé syndrome. There is no consensus on clinical surveillance; the recommendations given are provisional until a consensus conference is conducted.

* No routine screening is recommended for patients without signs/symptoms to avoid cumulative radiation exposure.

** Surveillance can start earlier in patients with a family history of renal tumor before age 30 years.

1. Sattler E, et al. 2006 Feb 27 [Updated 2020 Jan 30]. In: GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2024. https://www.ncbi.nlm.nih.gov/books/NBK1522/.

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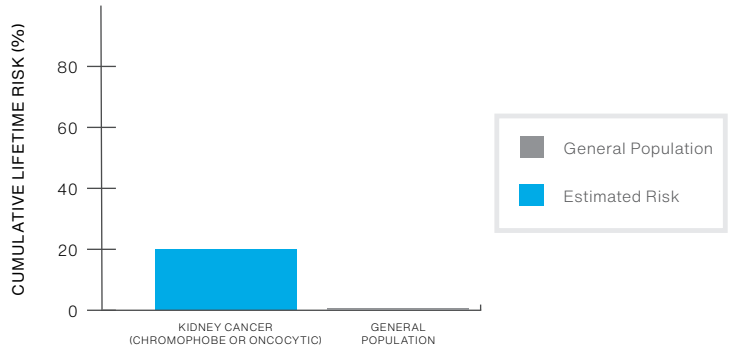
Understanding Your Positive *FLCN* Genetic Test Result

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

5 Things to know

1	*FLCN* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *FLCN* gene.
2	Birt-Hogg-Dubé syndrome	People with *FLCN* mutations have Birt-Hogg-Dubé syndrome (BHDS).
3	Cancer risks and other medical concerns	You have an increased chance to develop kidney (renal) cancer and multiple benign (non-cancerous) skin tumors. You also have an increased chance to develop multiple lung cysts which can cause too much air to get in between the lung and chest wall, causing lung collapse (pneumothorax).
4	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.
5	Family	Family members may also be at risk – they can be tested for the *FLCN* 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.

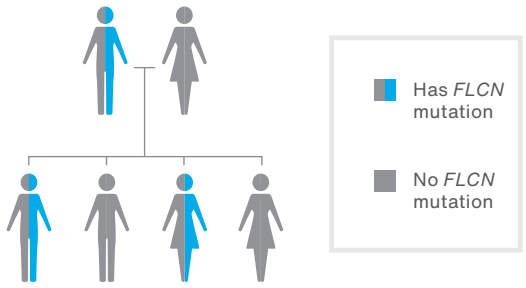
FLCN Mutation Lifetime Cancer Risks (%)*



* Because risk estimates vary in different studies, only approximate risks are given. Cancer risks will differ based on individual and family history.

FLCN Mutations in the Family

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



RESOURCES

- BHD Foundation bhdsyndrome.org
- National Society of Genetic Counselors nsgc.org
- Canadian Association of Genetic Counsellors cagc-accg.ca

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