

## Clinician Management Resource for Birt-Hogg-Dubé syndrome

This overview of clinical management guidelines is based on this patient's positive test result for a pathogenic or likely pathogenic *FLCN* variant. Unless otherwise stated, medical management guidelines used here are limited to those published in GeneReviews<sup>1</sup>. 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 <sup>1,^</sup>	AGE TO START	FREQUENCY
<b>Cutaneous manifestations</b>		
Detailed dermatologic exam.	Beginning at age 20 years	Individualized
<b>Lung cysts and/or pneumothorax</b>		
Discuss avoiding activities that might increase pneumothorax risk (e.g. working as a pilot, flying in unpressurized aircraft, diving).	At diagnosis	N/A
High-resolution CT or CT of the chest for visualization of pulmonary cysts and evidence for pneumothorax. Individuals with signs/symptoms of pneumothorax should immediately undergo chest x-ray and CT of the chest.	In adults at diagnosis	Only as needed*
<b>Renal tumors</b>		
Abdominal/pelvic MRI or abdominal/pelvic CT with contrast if MRI is not an option.	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.
<b>Counseling</b>		
Counsel patients to avoid the following agents/circumstances: cigarette smoking, high ambient pressures, radiation exposure	Individualized	Individualized
Genetic counseling by a medical geneticist, certified genetic counselor, or certified advanced genetic nurse to obtain a pedigree and inform affected individuals and their families regarding the nature, mode of inheritance, and implications of Birt-Hogg-Dubé syndrome to facilitate medical and personal decision making.	At diagnosis	Individualized

<sup>^</sup> The evaluations summarized in the table above are also recommended to establish the extent of disease and needs in an individual diagnosed with Birt-Hogg-Dubé syndrome, if not performed as part of the evaluation that led to the diagnosis.

\* 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 2024 Dec 5]. In: GeneReviews [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2025. <https://www.ncbi.nlm.nih.gov/books/NBK1522/>.

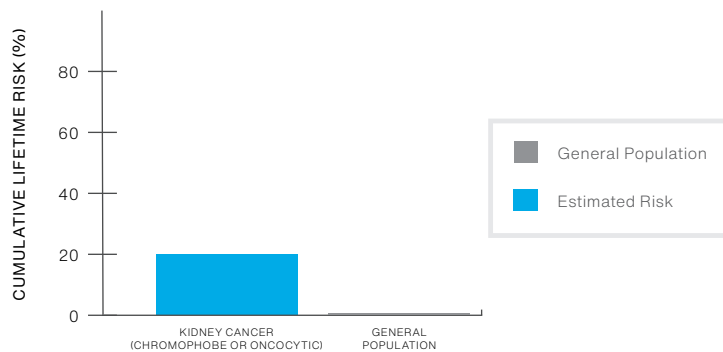
# Understanding Your Positive *FLCN* Genetic Test Result

## INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

### 5 Things to know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the <i>FLCN</i> gene.
2	Birt-Hogg-Dubé syndrome	People with a pathogenic or likely pathogenic <i>FLCN</i> variant 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 pathogenic or likely pathogenic <i>FLCN</i> 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.

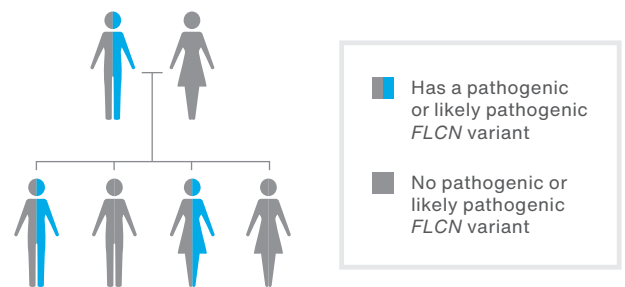
### *FLCN* Lifetime Cancer Risks (%)\*



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

### *FLCN* in the Family

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



### RESOURCES

- BHD Foundation [bhdysndrome.org](http://bhdysndrome.org)
- National Society of Genetic Counselors [nsgc.org](http://nsgc.org)
- Canadian Association of Genetic Counsellors [cagc-accg.ca](http://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.