BHD Foundation

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BIRT-HOGG-DUBÉ SYNDROME

Birt-Hogg-Dubé syndrome (BHD) is a rare genetic condition caused by a pathogenic variant in the FLCN (folliculin) gene.

BHD is associated with lung cysts, pneumothorax (also known as a punctured or collapsed lung), skin lesions and kidney cancer.

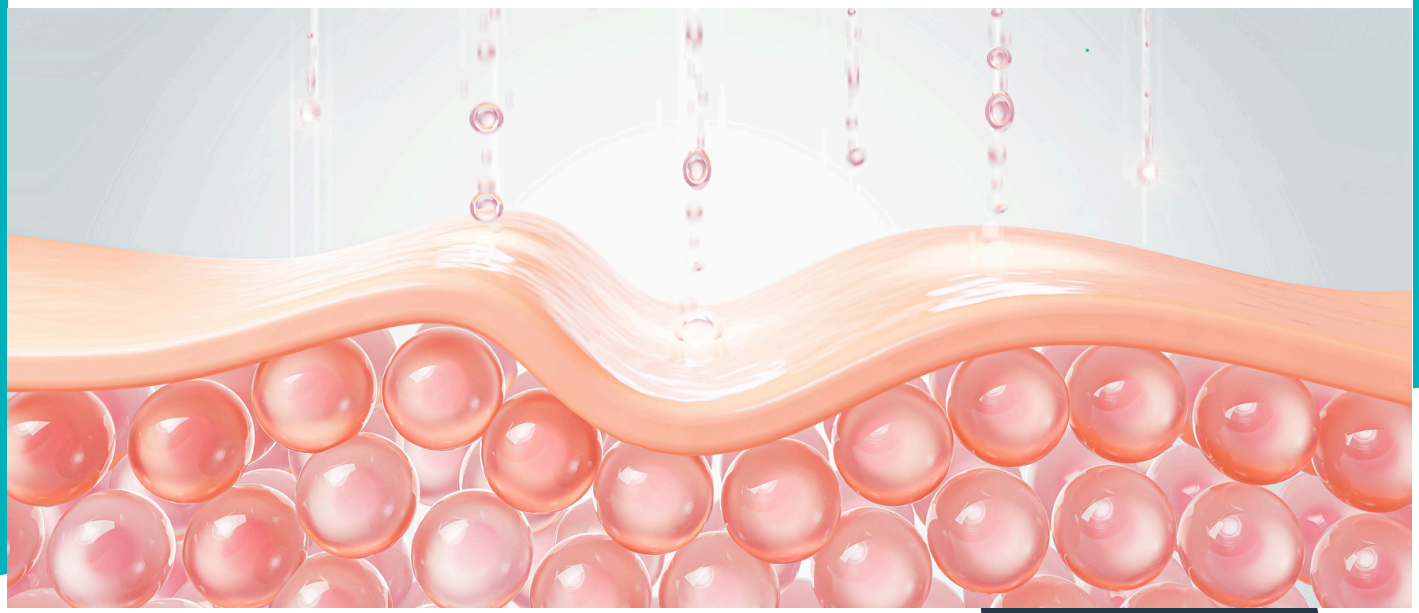
Early diagnosis of BHD is critical to ensure regular kidney surveillance to detect kidney cancer at an early stage.

The Skin

Cutaneous manifestations of BHD include fibrofolliculomas and trichodiscomas.

Fibrofolliculomas and trichodiscomas are 2-4 mm pale or flesh-coloured, smooth, dome-shaped papules that are mainly found on the face, neck and sometimes on the trunk; the age of onset is usually from 20 years. The papules are not painful or pruritic (itchy) but can be cosmetically undesirable.

Patients may have very few skin lesions or many, and they may increase in number over time. These skin lesions can confer a significant psychosocial burden, prompting patients to seek management from a dermatologist.

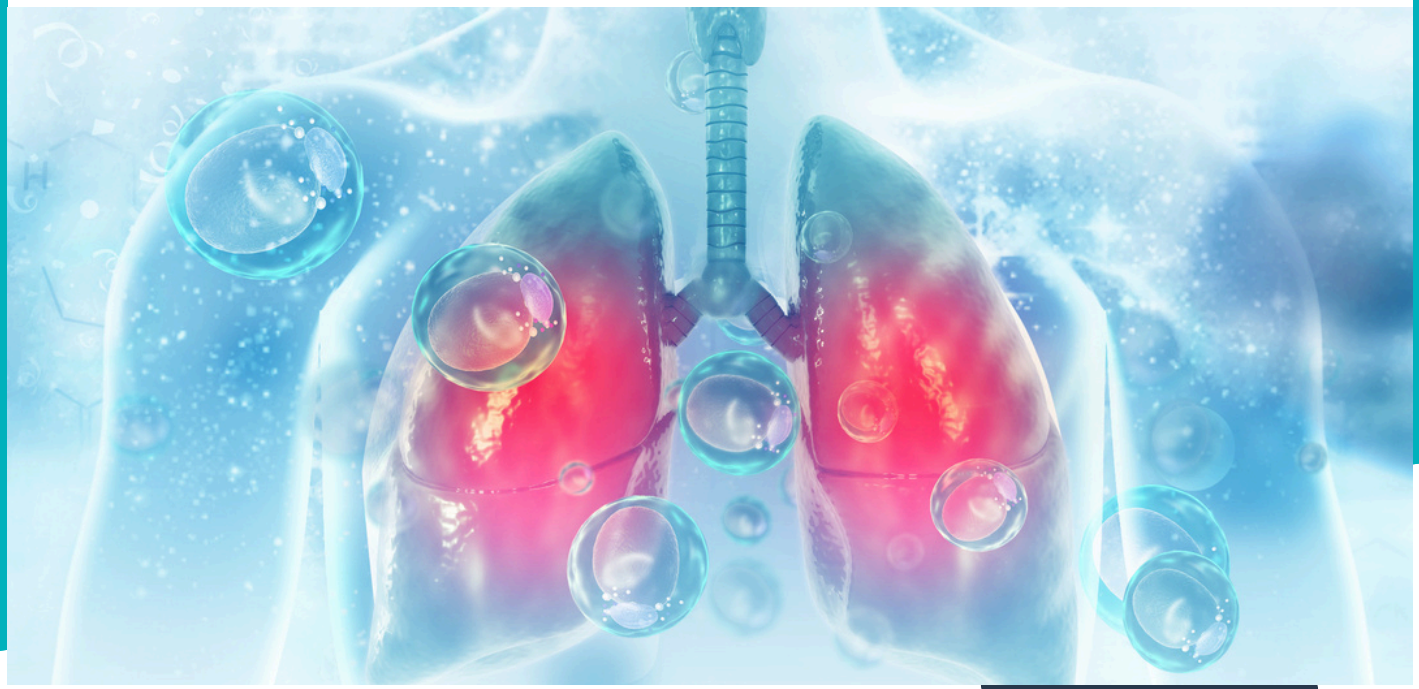


The Lungs

BHD is the most common genetic cause of familial spontaneous pneumothorax, which is often the first clinical presentation of BHD for patients in their 20s/30s.

If someone presents with recurrent spontaneous pneumothorax and/or a family history of pneumothorax, always consider BHD. Unless a BHD patient has a spontaneous pneumothorax, lung cysts are often an incidental finding on imaging. The most distinguishing feature of BHD-associated lung cysts is the basal predominance of the lesions.

The cysts can vary in size from less than 1cm to several centimetres. Lung cysts in BHD are round to lentiform (elliptical) in shape, lined by very thin walls, and commonly adjoin the pleural surface and blood vessels. The number of cysts in patients with BHD is variable, but most often patients have just a few cysts.



Diagnosis and Management

Diagnosis of BHD relies on a combination of clinical evaluation, family history (autosomal dominant inheritance pattern) and genetic testing. Genetic testing for mutations in the FLCN gene is the gold standard for diagnosis, so testing should be performed whenever possible and can confirm the diagnosis in 90% of cases.

If you suspect BHD, we recommend referring your patient for genetic testing at a local clinical genetics centre. Doctors or patients can also email us directly at contact@thebhdfoundation.org for advice on getting tested or finding a specialist.

There is currently no cure for BHD but with early diagnosis it is not normally life-threatening. Early diagnosis, regular surveillance for renal tumours and treatment of pneumothorax are major aspects to consider in the management of BHD.

Treatment of a rare condition like BHD should go beyond care of only the physical symptoms. Clinicians should be aware of and consider the psychosocial aspects of having a rare condition.

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THINK BHD SYNDROME

Does your patient have any of the following?

- Bilateral/multifocal renal tumours, especially hybrid or chromophobe subtype, or onset of renal tumours at a young age (50 years or less)
- History of BHD clinical manifestations within a family
- Dome-shaped papules (fibrofolliculomas)
- Spontaneous pneumothorax and / or lower lung zone predominant thin-walled cysts

If you answer YES to any of the above, it could be BHD.

Contact information

You can get in contact with us by using this email address:
contact@thebhdfoundation.org

References

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