



BIRT-HOGG-DUBÉ SYNDROME

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

BHD is associated with lung cysts, pneumothoraces, skin lesions and kidney cancer.

Early diagnosis of BHD is critical to ensure regular renal surveillance to minimise the risk of kidney cancer.

THE SKIN

Cutaneous manifestations of BHD include fibrofolliculomas, trichodiscomas and acrochordons (skin tags), of which fibrofolliculomas are the most common.¹

Fibrofolliculomas and trichodiscomas are 2–4 mm pale or flesh-coloured, smooth, dome-shaped papules that are mainly found on the face, neck and trunk; the age of onset is usually from 20 years.¹

Patients may have very few skin lesions or hundreds, and due to the genetic aspect of this condition, they may increase in number over time.²

Photos show variation in number and size of fibrofolliculomas in four different people.



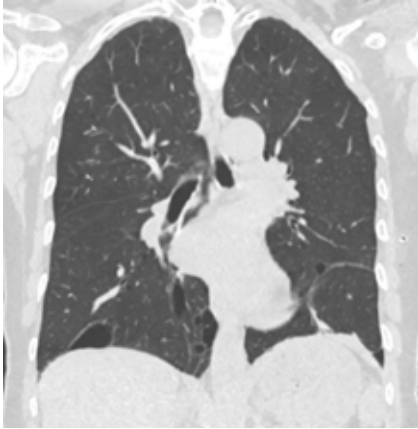
REMEMBER:
Early diagnosis of BHD is critical to ensure regular renal surveillance to minimise the risk of kidney cancer.

Permission for publication of images was given by members of the BHD community.

THE LUNGS

BHD is the most common known genetic cause of familial pneumothorax, which is often the first symptom reported in a BHD patient in their 20s/30s .

If someone presents with recurrent or a family history of pneumothoraces, always consider BHD.⁵



Coronal CT of a patient with BHD syndrome who had undergone a left-sided pleurectomy.⁷

Reproduced with permission from Nikolić and Marciniak.

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.⁶

The cysts can vary in size from less than 1cm to several centimetres, and are irregular in shape but usually have visible, thin, uniform walls. The number of cysts found in BHD patients ranges from just a few to more than 400.

THE KIDNEYS

The most life-threatening complication of BHD is renal cell carcinoma, which occurs in about 25% of patients at an average age of 50 years (can occur in young people).³ They can be bilateral and/or multifocal and can reoccur.

Histologically, they are typically chromophobe, oncocytoma or a hybrid of the two, however clear cell carcinoma and papillary carcinoma have been reported.⁴

These tumours are normally slow-growing, therefore regular monitoring and removal once they reach 3 cm is critical to reduce the risk of metastatic disease.

DIAGNOSIS

There are no current, official diagnostic guidelines for BHD. 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 should be performed where possible and can confirm over 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@bhdsyndrome.org for advice on getting tested or finding a specialist.



Scan the QR code for a list of clinic genetic services.

MANAGEMENT

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 pneumothoraces are major aspects to consider in the management of BHD.⁸

However, 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.

Doctors or patients can email us directly at contact@bhdsyndrome.org for further information about BHD, specialist care and support.



THINK **BHD** SYNDROME

Does your patient have any
of the following?

- B** Bilateral/multifocal renal tumors or young age of onset of renal tumors
- H** History within family
- D** Dome-shaped papules (fibrofolliculomas)
- S** Spontaneous pneumothorax and/or predominantly basal cysts

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



CONTACT US

To find out more about BHD including further details on management:

Email us at contact@bhdsyndrome.org

Visit our website at www.bhdsyndrome.org



Access our BHD explainer video, clinician resources, patient resources, references and more through our QR code.



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