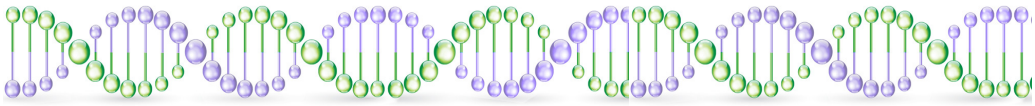
Information Sheet



Birt Hogg Dubé syndrome and the FLCN gene

Birt Hogg Dubé syndrome (BHD) is an inherited syndrome caused by a mutation in the Folliculin gene (FLCN). It is very rare, with around 400 families known worldwide.

How is Birt Hogg Dubé syndrome diagnosed?

The diagnosis of Birt Hogg Dubé syndrome is usually based on clinical criteria, with genetic testing of the FLCN performed to confirm the diagnosis and allow predictive testing of blood relatives.

Individuals with Birt Hogg Dubé syndrome almost always develop small skin coloured lumps on the face and chest (benign skin tumours called fibrofolliculoma) which appear in the 20s. (You can see some examples on our website.) Fibrofolliculoma are rare and all individuals with a biopsy-proven fibrofolliculoma should consider genetic testing.

Individuals with Birt Hogg Dubé syndrome have a high chance of developing cysts in the lungs. These may occur at a young age and may cause the lung to collapse (spontaneous pneumothorax). When these cysts occur at the base of the lung, and cannot be explained by other causes such as smoking, there is a 30% chance of an FLCN mutation.

Kidney tumours occur in 20 to 30% of individuals with Birt Hogg Dubé syndrome. These tend to be bilateral, multifocal and slow growing. In some cases, these tumours may develop into cancers. The tumours are usually an unusual type such as an oncocytoma or a chromophobe tumour. All individuals with an oncocytoma or a chromophobe tumour diagnosed before age 50 should consider genetic testing for Birt Hogg Dubé syndrome.

How is Birt Hogg Dubé syndrome managed?

The severity of the signs and symptoms vary among affected individuals, even within the same family, and is not predictable.

- Screening for renal tumours should start at age 20 with an abdominal MRI, followed by annual high quality renal ultrasound or MRI every 2 to 3 years.
- Because of the risk of spontaneous pneumothorax (a "collapsed lung"), a MedicAlert bracelet should be worn. Smoking and scuba diving (due to high ambient pressures) should be avoided.

Is Birt Hogg Dubé syndrome inherited?

Yes. Birt Hogg Dubé syndrome is a hereditary cancer syndrome caused by a mutation in the FLCN gene. There is a 50% chance of a person who carries a germline FLCN mutation, whether male or female, passing the mutation to their son or daughter. If a mutation is identified, then predictive testing is available for blood relatives.

Does this sound like you or your family?

Have you been diagnosed with a fibrofolliculoma or has an FLCN mutation been detected in a blood relative? Genetic testing is available. Make an appointment with Dr Hilda High at Sydney Cancer Genetics. It is a confidential opportunity to discuss your personal and family history of cancer and genetic testing can be organised, if needed.

More information is available on our website, including links to the following:

- We haven't found a specific Australian Support Group for this syndrome yet. However, BHD Foundation is based in London and has good resources.
- The Cancer Genetics section of the Cancer Institute's eviQ website provides up-to-date Australian-based management guidelines
- The US National Library of Medicine website has more information about this syndrome.

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