Birt-Hogg-Dubé Syndrome

Information for patients

What is Birt-Hogg-Dubé (BHD) Syndrome

BHD is a rare inherited condition. People with BHD may have benign (harmless) skin bumps, called “fibrofolliculomas” which are typically seen on the head, neck and upper trunk (torso). People with BHD can also develop air-filled cysts in their lungs, which sometimes lead to lung collapse (pneumothorax). They also have an increased risk of kidney cancer.

Is BHD syndrome inherited?

A change in a specific gene called FLCN, which creates a protein called folliculin, is known to cause BHD. As BHD is a dominantly inherited genetic condition, this means if you carry a change in your FLCN gene, your children and possibly your siblings would have a 1 in 2 (or 50%) chance of also carrying the same change.

How is BHD diagnosed?

BHD is commonly diagnosed with a genetic test. This is normally done by taking a blood or saliva sample to test for alterations in the FLCN gene. However, a small proportion of people may be diagnosed with BHD based on clinical grounds (their symptoms and medical signs) even if the genetic test does not show a fault in the folliculin gene. This is because the genetic test is not able to pick up every type of genetic alteration that can cause BHD.

If the genetic alteration has been identified in the family, then we can offer testing to other family members to determine if they are affected or not. However, if we have not been able to identify the genetic cause but still strongly suspect someone is affected by BHD based on clinical grounds then other family members will require testing and monitoring over time to determine if they are also affected.

Management of BHD

Renal Cancer – Around 15-30% of people with BHD develop renal (kidney) cancer. For this reason, individuals with this condition are advised to have a renal ultrasound scan on an annual basis from aged 18. If an abnormality or lump is seen on the kidney, further imaging with an MRI may be needed to help determine if the lump is cancerous. Some centres use MRI scans for screening but in South East Scotland, evidence shows that an ultrasound scan is adequate for detecting early renal cancer in individuals with BHD.

Pneumothorax - Individuals with BHD can develop lung cysts which on rare occasions can leak resulting in a pneumothorax (air trapped in the space surrounding the lungs/collapsed lung). This can lead to breathlessness and chest pain. Current guidelines recommend considering a CT scan following diagnosis to look for cysts. If cysts are diagnosed, we will then refer you to the respiratory doctors for further advice. However, it is important to realise that many people with lung cysts due to BHD will not develop a pneumothorax.

You should however avoid cigarette smoking and be aware of activities involving high ambient pressures such as scuba diving or flying in unpressurised aircraft, which increase the risk of pneumothorax.

The respiratory team may also undertake some further investigations to assess how the lungs are working.

Skin - Sunscreen may be helpful to protect against the development of BHD skin fibrofolliculomas, but more studies are needed to confirm its effectiveness. We also ask people with BHD to be vigilant about their skin and have a low threshold to seek medical advice if they notice any changes in their skin lesions such as pain, discomfort or change in size.

Bowel Cancer - If there is a family history of bowel cancer, then you may also be offered a colonoscopy.

The role of the clinical genetics service

The role of the genetics service is to offer BHD testing, to inform you about the condition and to organise your initial scans. We ask your GP to organise subsequent renal scans. In the clinic we also look at your family history to decide who else in your family might need testing for BHD. Letters are provided for you to pass onto your relatives allowing them to access genetic testing. We are also able to discuss reproductive options for future family planning.

More information

The BHD Foundation website provides a wide range of resources to inform, empower and support people with BHD:

https://bhdsyndrome.org

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Contact information

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