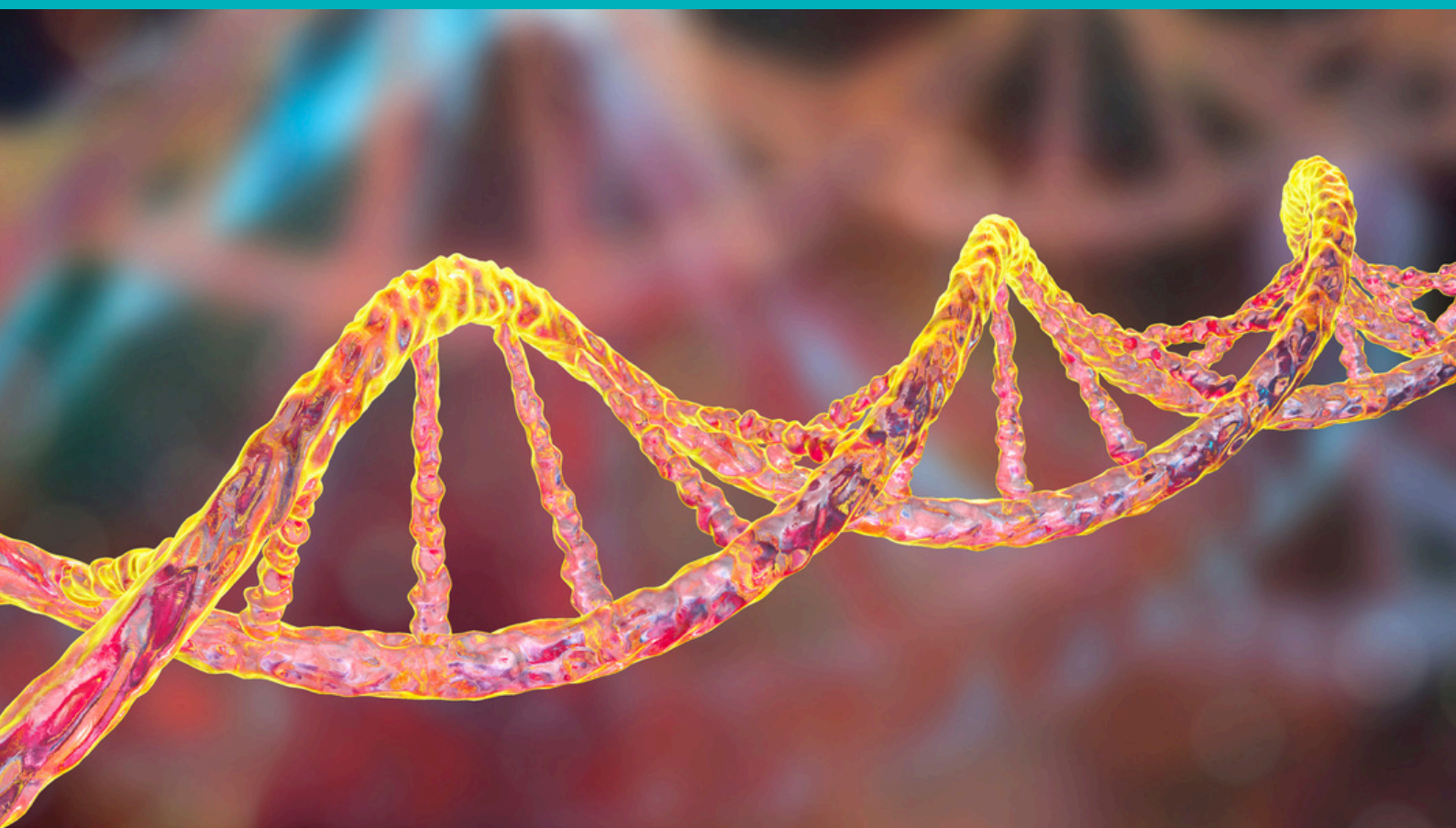


Introduction to BHD



What is Birt-Hogg-Dubé syndrome?

Birt-Hogg-Dubé Syndrome (BHD) is a rare genetic condition caused by mutations in the folliculin gene.

People with BHD can get:

- Skin bumps (called fibrofolliculomas - mostly on the face, neck and upper body)
- Lung Cysts
- Collapsed lung (pneumothorax)
- Kidney cancer (renal cell carcinoma)

BHD affects people differently. If you have BHD you may have none, some, or all of these symptoms.



Who should get tested?

1. Do you have any pale skin bumps on your face, ears, neck or upper body?
2. Have you ever had a collapsed lung, or been told you have lung cysts?
3. Have you ever had more than one kidney cyst or tumour?
4. Does anyone in your family have BHD or any of these symptoms?

If the answer is yes to one or more of these questions, you should talk to your doctor about BHD.

Testing is important so you can monitor your health and inform any family members so that they can also be tested.



How does gene testing work?

- The only way to know if you have BHD is by getting a genetic test.
- A small sample of blood is taken, DNA is extracted from blood cells, and the folliculin gene is checked to look for a mutation.
- Your doctor should be able to help arrange this for you.
- You may also want to see a genetic counsellor to support you and your family. Information on genetic counselling can be found on our website.



Symptoms: Skin

Fibrofolliculomas and Trichodiscomas

- Fibrofolliculomas and Trichodiscomas are small skin bumps most commonly found on the face, neck and upper body in people with BHD. To the naked eye they appear identical, but differences may be seen when looked at under a microscope. More research is needed to help understand these differences.
- 9 in 10 people with BHD will get these skin bumps. They usually start to appear from the age of 20. You may develop more as you age.
- You may have just a few, or hundreds of these bumps. They usually start to appear in your twenties and you may develop more as you age.
- They are not cancerous or dangerous to your health. Some people living with BHD feel self-conscious about them. You can seek help from a dermatologist if these skin bumps are affecting you.

Examples of fibrofolliculomas:



Photos courtesy of Dr Derek Lim, University of Birmingham, UK

Treatment options

- Several treatment options for fibrofolliculomas are available, however these are temporary and do not prevent the growth of new fibrofolliculomas.
- Some treatments may lead to scarring, and you should discuss any options carefully with your doctor and let them know you have BHD.
- Counselling may also be helpful if you are self-conscious about your skin. Available treatments for fibrofolliculomas include:

1. Laser, cryoablation (freezing technique)
2. Radiofrequency ablation (use of radiowaves)
3. Hyfreaction (uses electricity)
4. Cautery (can be hot or cold treatment)
5. Curettage (often combined with another technique) or surgery



Symptoms: Lung

Lung cysts

- Lung cysts are sacs of air that form on the surface of the lung.
- 8 in 10 people with BHD get lung cysts.
- The number and size of lung cysts varies from person to person and are best seen by CT scans. They do not normally affect how well your lungs function. Lung cysts may rupture and can cause collapsed lungs in BHD patients.

Collapsed lung

Around 1 in 4 people with BHD will experience at least one episode of a collapsed lung (also called spontaneous pneumothorax).

Symptoms include:

- a sharp stabbing pain on one side of the chest
- the pain is made worse by breathing in
- difficulty breathing/breathlessness

They are not normally life-threatening if you receive treatment, or if the collapse is very small.

It is therefore important to be able to recognise the symptoms of a collapsed lung so you can get an x-ray and seek treatment quickly. It can be useful to take a clinical description of BHD with you, as well as any medical history describing any lung treatments you have had previously.

Treatments available for collapsed lung

The treatment you receive for a pneumothorax will depend on a number of factors and you should always discuss available options and their side effects with your doctor.

You should always inform your doctor you have BHD and whether you have had any previous surgery on your lungs as this may effect which treatment you receive.

Available treatments for pneumothorax include:

- Chest Thoracostomy: a plastic tube is inserted into the chest to allow the air to drain and the lung to reinflate naturally
- Pleurodesis: a procedure that sticks the outside surface of the lung to the chest wall preventing further collapse. Pleurectomy (removal of the lining between the chest wall and the lung) can also be performed making it easier for the the lung to stick to the chest wall.

Surgery to remove a part of the lung (resection) or the cysts (blebectomy or bullectomy) can also be performed but it is important to discuss this fully with your surgeon as you may experience decreased lung function following the surgery.

Symptoms: Kidney

BHD is associated with an increased risk of kidney cysts and tumours. Up to 1 in 3 people with BHD will get kidney cancer. It is recommended that BHD patients have regular abdominal imaging scans to detect any cancer as early as possible. Kidney tumours associated with BHD are often multifocal and can occur in both kidneys but are normally slow growing, rarely spread and not aggressive. The most common symptoms of kidney cancer include:

- Blood in your urine
- Pain in your back or side – this can vary from a sharp stabbing to persistent dull ache
- A lump or swelling in your side

Other symptoms of kidney cancer include:

- Extreme tiredness,
- Unexplained weight loss
- High blood pressure (which can also be a risk factor for kidney cancer)
- A fever and/or night sweats



- BHD is not normally life-threatening if managed properly, most people with BHD lead normal lives.
- You should get regular kidney scans and learn the symptoms of a collapsed lung, so you know when you need to see a doctor.
- Finding a doctor who knows about BHD is important so you receive the best care.
- There is a Birt-Hogg-Dubé patient support group on Facebook. This is a very active community where you can ask questions, express concerns or share your experiences of having BHD, including recommendations of doctors.
- The BHD Foundation can also support you in finding a specialist doctor in your area and provide help with getting a genetic test.
- We also have weekly research blogs, a monthly newsletter, and run events to connect BHD patients with researchers and clinicians.

Contact us:

If you would like to get in touch with the BHD Foundation, please email our team: contact@bhdsyndrome.org

For more information about our work and BHD, you can visit our website: www.thebhdfoundation.org