

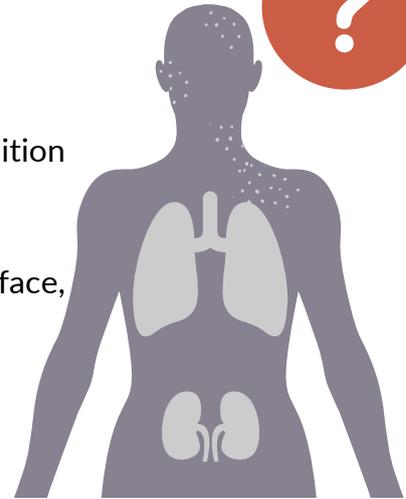
Introduction to BHD

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.

The BHD foundation is here to support you throughout your BHD journey .

Please visit our website for more information, or get in touch with us by email or twitter.



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FIBROFOLLICULOMAS

- Fibrofolliculomas are small pale skin bumps most commonly found on the face, neck and upper body in people with BHD.
- 9 in 10 people with BHD get fibrofolliculomas.
- 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**, but you may feel self-conscious about them.
- It is important to see a dermatologist because there are a number of skin conditions that may look similar - they can tell you what type of skin bumps you have.

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:
Laser, cryoablation (freezing technique), radiofrequency ablation (use of radiowaves), hyfreaction (uses electricity), cautery (can be hot or cold treatment), curettage (often combined with another technique) or surgery.

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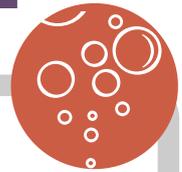
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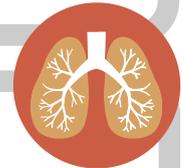
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Symptoms: Lung



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/shortness of breath
- 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 FOR COLLAPSED LUNG

- Several treatment options are available, however these are not specific to BHD.
- You should let your doctor know you have BHD as this may affect which treatment you receive.
- Available treatments 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.

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Symptoms: Kidney



KIDNEY TUMOURS

- 1 in 3 people with BHD get kidney cancer.
- In most cases, the tumours are generally **slow growing, rarely spread and are not aggressive** and with routine screening they are not usually life-threatening.
- It is **strongly recommended for BHD patients to have routine scans** to check for kidney tumours and to monitor their growth. Tumours are normally removed when they reach 3 cm.
- As BHD is a genetic disease and more tumours may develop over time, it is very important to tell your doctor you have BHD if you are having kidney surgery to preserve as much of the healthy kidney as possible.
- The most **common symptoms of kidney cancer** include:
 - blood in your urine
 - pain below your ribs
 - a lump or swelling in your side
 - unexplained weight loss
- If you have any of these symptoms you should contact your doctor immediately.



WHAT NOW?

- 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.

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