

BIRT-HOGG-DUBÉ SYNDROME

WHAT ARE THE AIMS OF THIS LEAFLET?

This leaflet has been written to help you understand more about Birt-Hogg-Dubé syndrome (BHD). It tells you what it is, what causes it, what can be done about it, and where you can find out more about it.

WHAT IS BIRT-HOGG-DUBÉ SYNDROME?

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

WHAT CAUSES BIRT-HOGG-DUBÉ SYNDROME?

BHD is caused by a fault in a piece of DNA which is a gene called “folliculin”.

IS BIRT-HOGG-DUBÉ SYNDROME HEREDITARY?

Yes, it can be passed down from one generation to the next. There is normally a family history of BHD. Clinical signs of BHD may differ between people even within the same family.

WHAT ARE THE SIGNS OF BIRT-HOGG-DUBÉ SYNDROME?

There are several different clinical signs associated with BHD. The signs may vary between people and may go unnoticed for many years.

Many studies have looked at how common the signs of BHD are.

It is thought that:

- Around 9 in 10 people get skin bumps (fibrofolliculomas or trichodiscomas)
- Around 8 in 10 people get lung cysts
- Around 1 in 4 people get collapsed lungs
- Around 1 in 3 people get kidney cancer.

The skin and lung signs seen in BHD typically occur when a person is in their 20s and 30s. People who develop kidney cancer are thought to do so around age 50 and therefore regular kidney scans are recommended.

Other skin signs have also been reported. These include:

- Skin tags (Acrochordons)
- Additional types of small bumps on the face (Angiofibromas)
- Lumps underneath the skin (Lipomas)
- Small bumps on the inside of the lips (Oral papules)

Some patients may not have any signs, or develop them later in life.

WHAT DOES BIRT-HOGG-DUBÉ SYNDROME LOOK LIKE?

BHD skin signs include skin-coloured bumps (fibrofolliculomas and trichodiscomas) typically 2-5 mm across. These skin bumps appear primarily on the head, neck and upper trunk and are not cancerous. A person may have just a few or may have hundreds. A person with BHD tends to get more skin bumps as they age.

HOW IS BIRT-HOGG-DUBÉ SYNDROME DIAGNOSED?

BHD is commonly diagnosed with a genetic test. This is normally done by taking a blood or saliva sample to test for disease causing genetic changes in the folliculin gene. However, a small proportion of people may be diagnosed on clinical grounds with BHD even though the genetic test does not show a fault in the folliculin gene.

CAN BIRT-HOGG-DUBÉ SYNDROME BE CURED?

No, there is currently no cure for BHD. However, there are kidney screening tests and treatments to help patients with BHD and with this regular care, most people with BHD live a normal life.

HOW CAN BIRT-HOGG-DUBÉ SYNDROME BE TREATED?

Skin bumps in BHD such as fibrofolliculomas and trichodiscomas are not dangerous. BHD patients may want to have them removed and this may be done using minor surgical techniques under local anaesthetic. BHD skin bumps tend to grow back so surgical treatments will need to be repeated from time to time.

The treatment of collapsed lungs in BHD is similar to how a collapsed lung is treated generally. Because collapsed lungs often recur in BHD patients, doctors may recommend a procedure called "pleurodesis". This procedure involves sticking the lung to the chest wall internally and should help prevent the lung from collapsing again.

To identify and treat any kidney cancer as early as possible, patients should have regular kidney scans typically from the age of 40. Surgery to remove any tumours is normally recommended when the largest tumour reaches 3 cm.

SELF-CARE (WHAT CAN I DO?)

It is important to be aware of the symptoms of a collapsed lung, which may include

chest pain and sudden breathlessness, so medical help can be sought as soon as this happens. Sudden changes in air pressure can increase the chances of developing a collapsed lung, so avoid activities where there will be large changes in air pressure (such as flying in unpressurised planes or scuba diving). You should consult your doctor before taking part in these activities. You must not fly if you have any symptoms of a collapsed lung.

Sunscreen may be helpful to protect against the development of BHD skin bumps, but more studies are needed to confirm its effectiveness.

WHERE CAN I GET MORE INFORMATION ABOUT BIRT-HOGG-DUBÉ SYNDROME?

Links to other Internet sites:

The BHD Foundation was established by the Myrovlytis Trust to support the BHD community and foster relationships between patients, clinicians and researchers.

The BHD Foundation website (www.thebhdfoundation.org) provides a wide range of resources to inform, empower and support people with BHD. This includes interviews with the BHD community, blog posts explaining the latest research and information leaflets (see below). The BHD Foundation also holds regular 'Meet the Expert' events, biannual community-focused symposiums and a monthly newsletter.

The BHD Foundation can help you to locate your nearest BHD specialist and will answer any questions about BHD via email (contact@thebhdfoundation.org).

The BHD Syndrome International Registry (BIRT) was launched in March 2022 with the aim of driving forward research to gain new insights into BHD. Find out more about the BIRT registry at <https://birt.healthie.net>.



Web links to detailed leaflets:

BHD Foundation Leaflets:

<https://www.thebhdfoundation.org/living-with-bhd/resources>

Links to patient support groups:

BHD Patient Facebook Group:

<https://www.facebook.com/groups/454615147912181/>

For more resources from the BHD foundation, scan the QR code



Please note that the BAD provides web links to additional resources to help people access a range of information about their treatment or skin condition. The views expressed in these external resources may not be shared by the BAD or its members. The BAD has no control of and does not endorse the content of external links.

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

**BRITISH ASSOCIATION OF
DERMATOLOGISTS**

PATIENT INFORMATION LEAFLET

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