



[www.BHDSyndrome.org](http://www.BHDSyndrome.org)

## Diagnosis Information

This pamphlet tells you how to get tested for Birt-Hogg-Dubé syndrome and what to do if you have it.

## Birt-Hogg-Dubé Syndrome

Birt-Hogg-Dubé (BHD) Syndrome is a rare disease caused by changes in the Folliculin gene.

People with BHD can get:

1. Skin bumps (fibrofolliculomas)
2. Lung cysts
3. Collapsed lungs (pneumothorax)
4. Kidney cancer (renal cell carcinoma)

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

## Who should get tested for BHD?

If the answer is yes to any of these questions, you should talk to your doctor about BHD syndrome.

1. Do you have any pale skin bumps on your face, ears, neck or upper body?
2. Do you have chest pains and feel short of breath?
3. Have you ever had one or more kidney cysts or tumours?
4. Does anyone in your family have BHD or any of these symptoms?

## Why should I get tested?

BHD is caused by mutations in the Folliculin gene.

**The only way to know if you have BHD is to have a gene test.**

If you have BHD, your family members know to get tested too. When your mutation is known, testing is quicker and cheaper for relatives.

### How does gene testing work?

A small sample of your blood will be sent to a lab. They will test the DNA in your blood cells to see if you have BHD.

Your doctor should be able to arrange this for you.

### 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.

More information about BHD symptoms, treatment and support is available at [www.BHDSyndrome.org](http://www.BHDSyndrome.org)

### Who should I talk to?

1. You should find a doctor who knows about BHD and will find the best care for you.
2. BHD is a genetic condition, so you may want to talk to your Clinical Genetics Service. These are doctors and nurses with expert knowledge about genetic diseases, who will be able to help you and your family. Your doctor can organise this for you.
3. The BHD Foundation. We provide information about BHD to patients and their families, and can put you in touch with expert doctors. You can email us at [contact@BHDSyndrome.org](mailto:contact@BHDSyndrome.org)

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