

## What next?

Once you know you have BHD syndrome, you will know to monitor your health closely, you will be aware of symptoms to look out for, and will be able to react quickly and appropriately if you need medical attention.



You will probably want to find a doctor who is knowledgeable about BHD syndrome and will research the best care for you. The [Birt-Hogg-Dubé Syndrome: Clinical Introduction](#) pamphlet is written especially for medical professionals and you may find it helpful to share with your doctor. Since having Birt-Hogg-Dubé syndrome increases the risk of kidney tumours, you may want to ask your doctor to arrange regular kidney checks.

Further information on BHD symptoms and available treatments can be found in the other pamphlets in this series and on [www.BHDSyndrome.org](http://www.BHDSyndrome.org).

## Also available:

Birt-Hogg-Dubé Syndrome:  
Clinical Introduction

Birt-Hogg-Dubé Syndrome:  
Lung Symptoms and Treatment

Birt-Hogg-Dubé Syndrome:  
Skin Symptoms and Treatment

Birt-Hogg-Dubé Syndrome:  
Kidney Symptoms

Birt-Hogg-Dubé Syndrome:  
Kidney Treatment

## Birt-Hogg-Dubé Syndrome:

## Diagnosis Information



# BHD Birt-Hogg-Dubé Syndrome: Diagnosis Information

## Introduction

Birt-Hogg-Dubé (BHD) syndrome is a rare (1 in 200,000) genetic disorder caused by alterations in the gene *Folliculin*. BHD is characterised by the development of benign skin tumours (fibrofolliculomas), lung cysts that can cause collapsed lung (spontaneous pneumothorax), and kidney cancer (renal cell carcinoma).

BHD affects people differently. If you have BHD syndrome, you may have none, one, or all of the symptoms of BHD.

## Who should be tested for BHD?

These questions may help determine if you should suspect BHD syndrome:

### Fibrofolliculomas

- Do you have any whitish skin bumps on your face, ears, neck or upper body?

### Spontaneous pneumothorax

- Do you have chest pains and feel short of breath?

### Kidney cancer

- Have you ever had one or more kidney cysts or tumours?

### Family history

- Has anyone in your family been diagnosed with BHD syndrome or does a family member have any of the symptoms that characterise BHD?

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## Why should I be tested? How does it work?

The only definite way of knowing whether you have BHD syndrome is to look for mutations in your *Folliculin* gene.

If you suspect you may have BHD syndrome, you should talk to a doctor or genetic counsellor about pursuing a diagnosis.



He or she will usually arrange a blood test for you. A small sample of your blood will then be sent to a testing laboratory. The laboratory will analyse your DNA to determine if you have a *Folliculin* mutation.

Confirming you have BHD syndrome also allows you to alert family members to their risk.

Once the first person in a family has received a positive diagnosis, other family members can be tested more easily and at lower cost to see if they also have BHD syndrome.

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## Genetic Counselling

Knowing that you have a genetic condition in your family means that you have many things to consider, as well as taking care of your symptoms. You may want to think about meeting a member of the Clinical Genetics service.

The Clinical Genetics team which comprise of clinical geneticists (doctors specialised in diagnosing and management of genetic conditions) and genetic counsellors (trained individuals, usually with specialised graduate or nursing degrees). Their goal is to offer families an idea about what it means to have a genetic condition, outline local resources for clinical management of the condition, identify family members who are at risk and help find other relevant information.

A genetic counsellor will not necessarily be a specialist in BHD syndrome or rare kidney cancer, but they will be familiar with the overall issues and will be able to offer valuable guidance about how best you and your family can approach your situation.



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