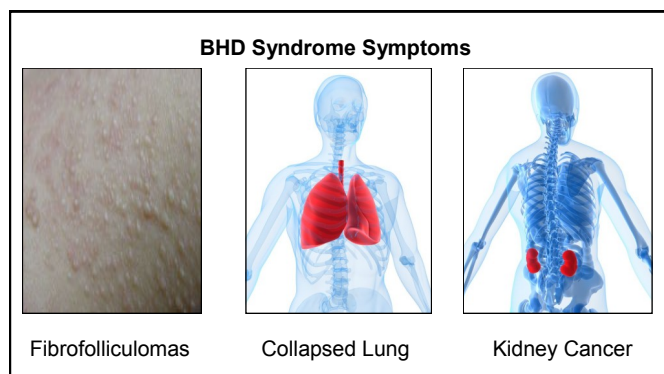


Birt-Hogg-Dubé Syndrome: Diagnosis Information

Introduction

Birt-Hogg-Dubé (BHD) syndrome is a rare genetic disorder characterised by the development of benign skin tumours, recurrent collapsed lung (pneumothorax) and, in some cases, kidney cancer.



There is no 'typical' BHD patient and individuals may show any, or all of these symptoms over time. BHD syndrome is caused by alterations (mutations) in the Folliculin gene. This means that BHD syndrome can be genetically diagnosed. In the UK, diagnosis is available through the NHS.

Who should be tested for BHD?

If you think you may have BHD, you should consult your GP. BHD is inherited, so if anyone in your family has BHD, you might also have it. Anyone closely related to someone with BHD, such as a parent, child or sibling, is particularly at risk, but more distant relations such as a grandparent, grandchild, aunt, uncle or cousin, might also have inherited the condition.

It is important to be diagnosed because once you know you have BHD, you will be more aware of changes in your health and subsequently respond to these changes appropriately e.g. if you feel you require medical attention.



In this respect a diagnosis will allow you to continue to lead a normal and healthy life.

Clinical Genetics

A BHD diagnosis will involve referral from your GP to a Medical Genetics Clinic. Specialist medical health care professionals will organise a genetic diagnosis for you, and will ask for information regarding your family history and possible routes of surveillance for yourself and other family members.

Confirmation that you have BHD Syndrome can lead to lots of questions and it is natural to have concerns after a positive diagnosis.

You will be able to discuss these concerns with either your GP, or with the clinical geneticists who handled your diagnosis - whoever you feel more comfortable with.

Clinical geneticists will probably not be specialists in the field of BHD Syndrome, but they will have been trained to deal with the issues arising from a positive diagnosis of a genetic disease such as BHD Syndrome.

They can help individuals explore what it means to have BHD syndrome on a personal level but also assist in discussing the wider implications of a positive diagnosis of a genetic disease. This may include family planning issues or help in identifying living family members who are at risk of having BHD Syndrome.

As a result you may want to consider:

- telling other family members, and letting them know how they too could be tested;
- if you have children, how having BHD syndrome could affect them, both emotionally and physically.

Your Medical Genetics Clinic will prepare and advise you on how to address these sensitive areas.

Also available in the series

[BHD Syndrome: Skin Symptoms & Treatment](#)

[BHD Syndrome: Lung Symptoms & Treatment](#)

www.BHDSyndrome.org