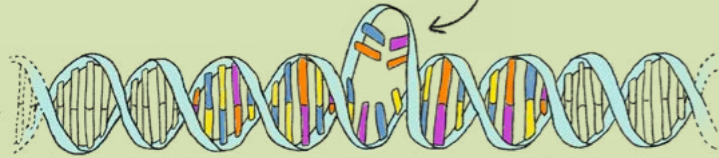


Information about Birt-Hogg-Dubé syndrome (BHD)

Birt-Hogg-Dubé (BHD) is a **rare hereditary syndrome**. A syndrome is a combination of conditions that arise from one cause. BHD is caused by a **DNA abnormality** in the **FLCN gene**.



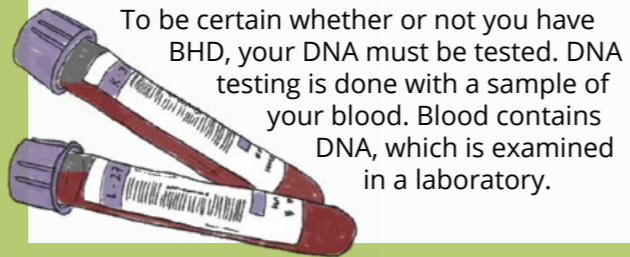
People with BHD often have non-cancerous **skin lesions** (fibrofolliculomas) and **cysts in the lungs** (air-filled sacs). In addition, there is an increased risk of a **collapsed lung** (pneumothorax) and **kidney cancer**.

How common is BHD?

We do not know exactly how common BHD is. In people with multiple **fibrofolliculomas** (bumps on the skin) there is a good chance that they have BHD. BHD is rarely found in people with kidney cancer or a collapsed lung.



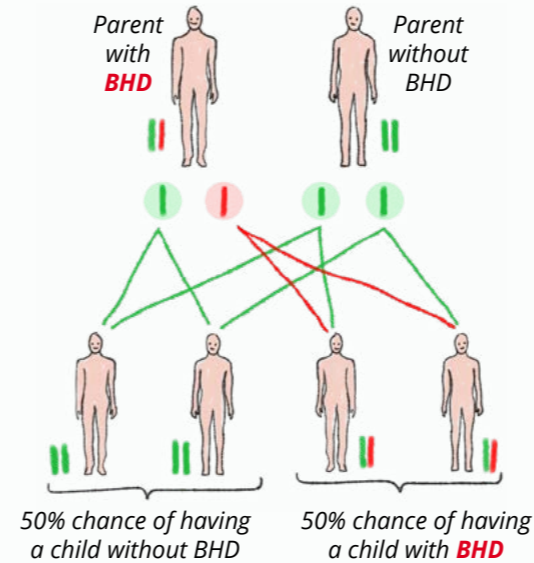
DNA test



To be certain whether or not you have BHD, your DNA must be tested. DNA testing is done with a sample of your blood. Blood contains DNA, which is examined in a laboratory.

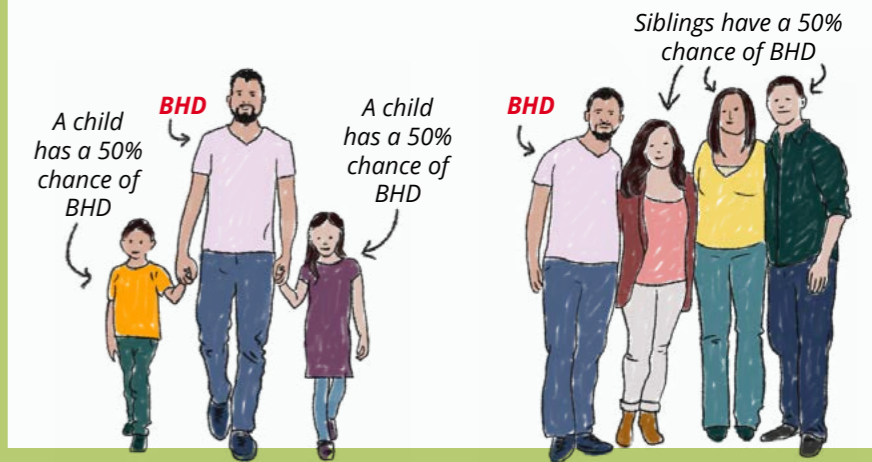
How is BHD inherited?

BHD is inherited in an **autosomal dominant** manner. Children of a parent with BHD each have a **50% chance (1 in 2)** of also having BHD. This applies to both sons and daughters.



What does this mean for family members?

Parents, children and siblings of people with BHD have a **50% chance** of also having this predisposition. For genetic advice and a DNA test, you can ask your GP to refer you to a clinical geneticist. DNA testing can be performed from (young) adulthood onwards.



What are the options if you want to have children?

More information about hereditary conditions when planning a family can be found at www.erfelijkheid.nl/kinderwens/wat-als-je-een-erfelijke-ziekte-kunt-doorgeven.

For tailor-made information, you can make an appointment with a **clinical geneticist**.



What complaints and symptoms might you experience?

Skin symptoms



Most people with BHD have skin symptoms. These are usually **fibrofolliculomas**: white bumps on the face, neck and upper torso.

Fibrofolliculomas are **non-cancerous**. Sometimes they are treated for cosmetic reasons.

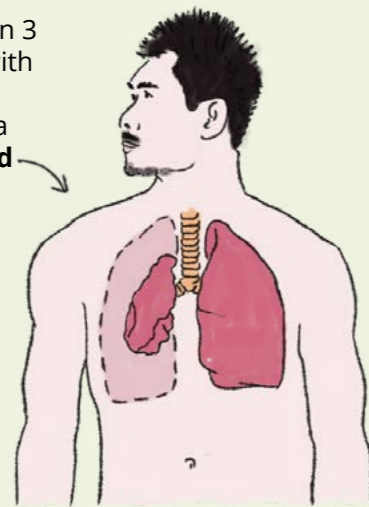


Pulmonary symptoms



Most people with BHD have multiple **lung cysts**. Lung cysts are air-filled sacs in the lungs. These cysts usually do not cause any complaints.

About 1 in 3 people with BHD will develop a **collapsed lung**.



This means that one lung collapses. This can lead to **shortness of breath**.

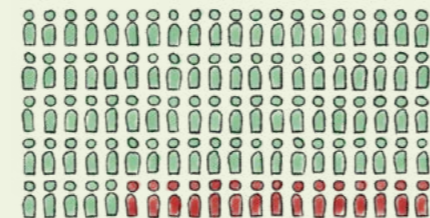
If you have BHD, but have never had a collapsed lung, it is not necessary to investigate whether you have lung cysts. We cannot prevent a collapsed lung.

If you have BHD and you do have a collapsed lung, we can try to prevent you from developing a collapsed lung again when treating the collapsed lung.

Kidney cancer



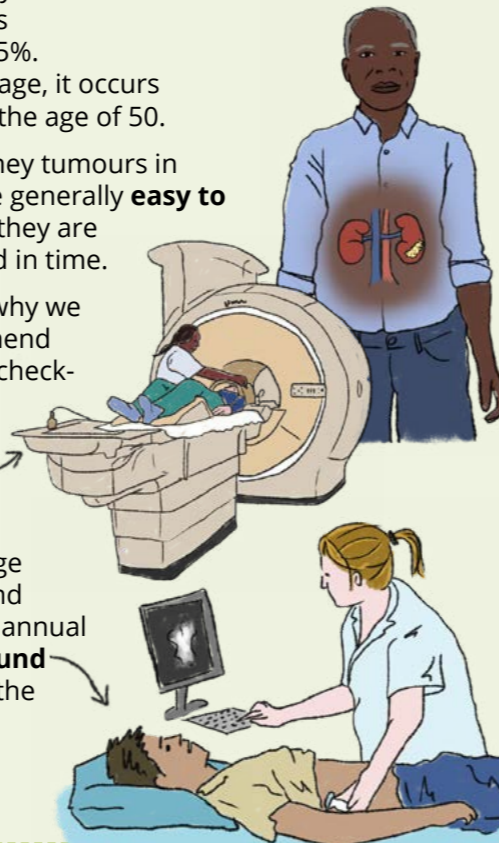
People with BHD have an increased risk of **kidney cancer**. The risk of kidney cancer is about 15%. On average, it occurs around the age of 50.



Of 100 people with BHD, 15 will develop kidney cancer

The kidney tumours in BHD are generally **easy to treat** if they are detected in time.

That is why we recommend regular check-ups: an **MRI scan** of the kidneys at the age of 20, and then an annual **ultrasound** scan of the kidneys via the GP.



How can you help?

- We advise you **not to smoke**. Smoking increases the risk of kidney cancer and of a collapsed lung.
- If you want to **dive**, ask a diving doctor or pulmonologist for advice. Diving is probably not wise for people with lung cysts.



- If you **fly** often (for example for your job), discuss this with a pulmonologist.



- If you need **general anaesthesia**, tell your doctor that you have BHD as the risk of a collapsed lung is higher.



Psychosocial consequences

People who discover that they have a hereditary condition can find this a **shock**. Social workers or psychologists also work at clinical genetics outpatient clinics. They can help you with questions, doubts and problems.



It is often assumed that having a hereditary condition has consequences for taking out **insurance** policies. This is rarely the case. More information about this can be found at www.erfelijkheid.nl/ziektes-en-dan/verzekeringen-en-erfelijke-ziektes. You can also discuss this with your clinical geneticist.

More information

www.erfelijkheid.nl (Dutch)
www.bhdsyndrome.org (English)
www.kanker.nl (Dutch)

