Your next steps

Find a specialist
All patients should be seen in a clinical genetics department or consultation.

Follow medical guidelines
Kidney surveillance every 1 to 2 years from age 20 years should be undertaken. Skin evaluation should be offered.

Monitor symptoms
Keep track of new or changing health issues

Consider treatment options
Discuss available therapies and potential surgical interventions.

Stay updated on Birt-Hogg-Dubé syndrome research
Clinical trials may offer new treatment possibilities.

Find a patient organisation
They can provide guidance and support.

Visit **www.genturis.eu** for surveillance guidelines

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

www.genturis.eu
E-mail: genturis@radboudumc.nl

European Reference Network

Funded by the European Union





Patient information

Birt-Hogg-Dubé syndrome

If you or a family member has been diagnosed with Birt-Hogg-Dubé syndrome, this guide provides clear medical information and practical next steps.

European Reference Network
for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

ERN GENTURIS
With every diagnosis we can help an entire family

What is Birt-Hogg-Dubé syndrome?

Birt-Hogg-Dubé syndrome is a rare inherited condition that increases the risk of developing kidney cancer. It is caused by disease-causing changes (pathogenic variants) in the *FLCN* gene.

Although BHD syndrome is considered rare, it is likely underdiagnosed because many affected individuals have mild or unrecognized symptoms. People with BHD syndrome may develop the following features:

- Skin changes: small, non-cancerous skin tumors called fibrofolliculomas, typically appearing on the face, neck, and upper chest from the third decade of life.
- Lung manifestations: Multiple lung cysts (small, air-filled spaces) are common. These usually do not affect breathing. However, in some people, a lung cyst can contribute to a collapsed lung (called a pneumothorax).
- Kidney tumours: Individuals with BHD syndrome have an increased lifetime risk (approximately 15–30%) of developing kidney tumors, which may be benign (non--cancerous) or malignant (cancerous). These tumors usually develop between 40 and 50 years of age.

How is Birt-Hogg-Dubé syndrome inherited?

BHD syndrome is inherited in an ***autosomal dominant pattern.*** This means that a person needs to inherit a disease-causing change in just one copy of the *FLCN* gene to develop the condition.

What causes Birt-Hogg-Dubé syndrome?

BHD syndrome results from disease-causing changes (pathogenic variant) in the *FLCN* (folliculin) gene. The *FLCN* gene normally helps regulate cell growth and function.

What are the diagnostic/ testing options?

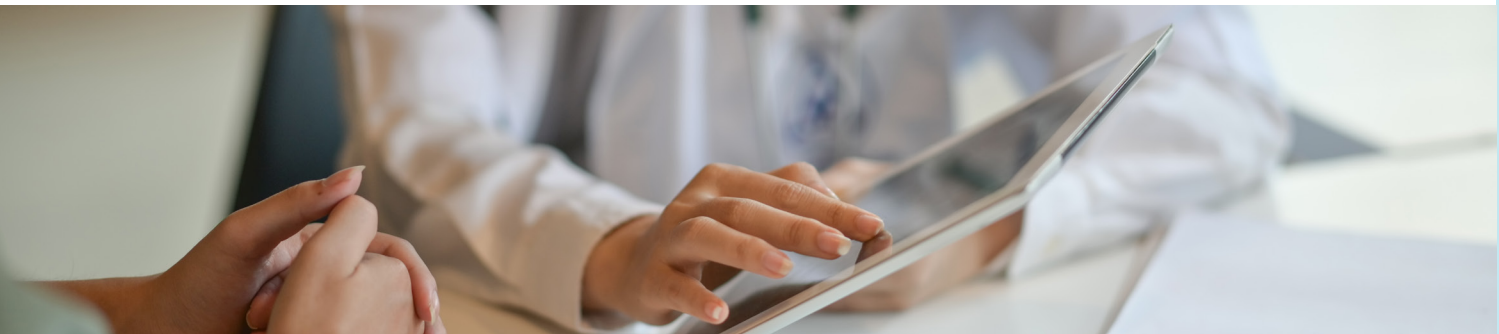
The diagnosis of BHD syndrome is based on a combination of clinical evaluation and genetic testing. Diagnostic options include:

- **Clinical assessment** of personal and family history, including kidney tumors, spontaneous pneumothorax, lung cysts, and characteristic skin lesions.
- **Physical examination**, particularly assessment of skin lesions by a dermatologist.
- **Genetic testing** of the *FLCN* gene to identify a pathogenic variant, which can be detected in most individuals.

What are the surveillance recommendations?

According to the ERN GENTURIS guidelines:

- Kidney: Every 1-2 years renal imaging preferable by MRI with contrast, otherwise ultrasound (from age 20 years and life--long).
- Lung: Regular chest imaging or lung function testing is not indicated. At diagnosis a baseline CT of the chest can be considered.
- Skin: Referral to dermatological evaluation and treatment at diagnosis or on request.



How this diagnosis affects you?

Receiving a genetic diagnosis requires careful medical supervision. Here are key points to consider:

How will Birt-Hogg-Dubé syndrome and the associated tumour risk impact your health?

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need a specialist evaluation over time.
- Since BHD syndrome is genetic, family members should also consider genetic testing and surveillance.

What medical steps should I take?

- **Step 1:** Schedule an appointment with a specialist in BHD syndrome.
- **Step 2:** Discuss options for genetic testing of relatives
- **Step 3:** Develop a personalised long-term surveillance plan with your doctor.

ERN GENTURIS provides information and resources to help you understand this condition and guide your medical team toward the best possible care.

More information on **Birt-Hogg-Dubé syndrome**: