

Clear cell

Although clear cell is one of the most common types of renal cell carcinoma, it is not common in families with BHD syndrome. Of the very small number of fatalities that have been associated with BHD, some were clear cell cases.

Testing

To determine the type of tumour, the tumour is first removed surgically and then a sample is sent to a pathology lab for testing. If the first result is not clear, you may want to ask that a section of your tumour be sent to a different lab for a second opinion.

The prognosis for BHD-related kidney cancer is generally positive. BHD-related cancers do not normally metastasise and so offer a good target for effective treatment. For more information on treatments, see the [Birt-Hogg-Dubé Syndrome: Kidney Treatment](#) pamphlet.

Most importantly, make sure that your kidneys are being regularly monitored by an expert clinical team.

Also available:

Birt-Hogg-Dubé Syndrome:
Clinical Introduction

Birt-Hogg-Dubé Syndrome:
Diagnosis Information

Birt-Hogg-Dubé Syndrome:
Lung Symptoms and Treatment

Birt-Hogg-Dubé Syndrome:
Skin Symptoms and Treatment

Birt-Hogg-Dubé Syndrome:
Kidney Treatment

Birt-Hogg-Dubé Syndrome:

Kidney Symptoms



BHD Birt-Hogg-Dubé Syndrome: Kidney Symptoms

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.

Kidney Symptoms

BHD syndrome increases the risk of cysts and tumours in the kidney. If you have BHD, it is important to have your kidneys monitored by regular scans every 1-2 years.

Kidney Cysts

Kidney cysts are rounded sacs usually filled with fluid.

- Simple kidney cysts are benign and are common in the older population. Treatment is only required if they affect the function of the kidney or another organ.
- Complex cysts can potentially be cancerous and may need to be removed. A complex cyst has a more irregular shape than a simple cyst, and other irregularities such as a thicker wall.

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Kidney Tumours

Kidney tumours associated with BHD syndrome are often multifocal and occur in both kidneys. They are detected by CT, ultrasound or MRI scans. Kidney tumours may be benign or cancerous.

Symptoms of kidney tumours can include the following:

- Blood in urine (haematuria).
- High blood pressure (hypertension).
- Weight loss and/or loss of appetite and/or fatigue (cachexia).
- Fever.
- Pain in the sides and/or lower back.
- Swelling (Oedema), especially of the legs and feet.
- Blood tests may show an elevated platelet count, an abnormal red blood cell count or elevated calcium levels.
- Some people experience night sweats (sleep hyperhidrosis).
- Sometimes the tumour may be palpable, i.e. the mass can be felt with your fingers.



It is only possible to be sure you have a kidney tumour by consulting a doctor, as all of these symptoms may be related to other conditions.

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Types of Kidney Tumours

Different types of kidney tumours grow at different rates, have different prognoses and react differently to treatment.

Oncocytomas

Oncocytomas are considered to be benign kidney tumours which can grow inside the kidney or on the surface. Though oncocytomas are benign, they may grow in places that affect blood flow in the kidney and so will need to be removed. This type of tumour does not spread (metastasise) to other parts of the body. About 3% of BHD-related kidney tumours are classified as oncocytic.

Mixed or Hybrid Oncocytic tumours

Hybrid tumours are a mixture of two types, oncocytic and another type, and are common among BHD-related tumours. 67% of BHD kidney tumours are hybrid.

Chromophobe

Chromophobe kidney cancer is often slow-growing, and does not appear to metastasise in people with BHD syndrome. About 23% of BHD-related kidney tumours are chromophobe.

Papillary

Although papillary renal cell carcinoma has been identified in BHD syndrome, it is rare.

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