Birt-Hogg-Dubé Syndrome is a rare, autosomal dominant disorder characterised by the development of benign epidermal tumours originating from hair follicles, pneumothorax and, in some cases, renal cancer.

There is no ‘typical’ presentation of symptoms as affected individuals may show any, or all of these symptoms over time.

BHD syndrome is caused by mutations in the Folliculin (FLCN) gene found on chromosome 17p11.2.

Pneumothorax

Spontaneous pneumothorax associated with BHD Syndrome are caused by the development of air filled blebs/cysts on the surface of the lung.

Symptoms can include acute chest pains, sudden shortness of breath, and an increased heart rate. There is no way to predict when lungs might collapse. In the general population, tall, thin, athletic males are more likely to have pneumothoraces than other people, but this does not necessarily apply to people with BHD.

Fibrofolliculomas

The fibrofolliculomas associated with BHD Syndrome develop as white bumps/growths on the skin of the head and upper torso. They often develop in the second or third decade of life, and an individual may develop none, one or several hundred over their lifetime. There is currently no preventative treatment.

Renal Cell Carcinoma

Chromophobe renal cancer and a mixed pattern of chromophobe and oncocytic renal tumours are typical for patients with BHD. However, other histological subtypes can occur, including clear-cell and papillary carcinoma, and several mixed patterns. Renal cancer is multifocal or bilateral in more than half of patients with BHD.

Previously, 27% of affected individuals were reported to have developed renal tumours (age of 31 – 74 years) but renal tumours have been diagnosed as early as age 20 years. Since the penetrance of RCC in BHD Syndrome is incomplete, the risk of renal cancer is not the same for all BHD patients. However, the unpredictability of its occurrence, and its serious nature, mean that the risk of RCC should be considered the same for all FLCN mutation carriers.

Appropriate clinical surveillance for BHD patients can be organised via a clinical geneticist or physician with an interest in BHD Syndrome.


Also available in the series
BHD Syndrome: Diagnosis Information
BHD Syndrome: Skin Symptoms & Treatment
BHD Syndrome: Lung Symptoms & Treatment

www.BHDSyndrome.org