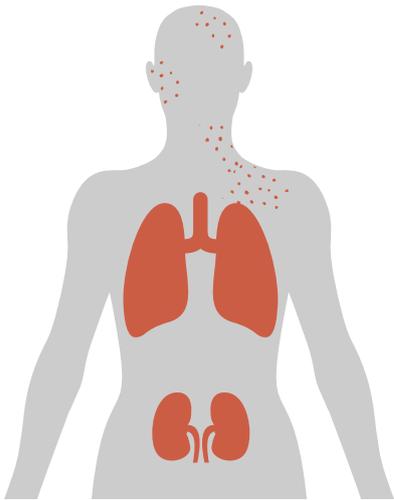


Birt-Hogg-Dubé Syndrome: Clinical Introduction

WHAT IS BHD?

Birt-Hogg-Dubé Syndrome (BHD) is a rare, autosomal dominant disorder caused by mutations in the gene *folliculin*.

BHD is characterised by fibrofolliculomas, lung cysts, pneumothorax and kidney tumours.



BE BHD AWARE!

- 9 in 10 people with BHD get **skin lesions**
- 8 in 10 people with BHD get **lung cysts**
- 1 in 4 people with BHD get **pneumothorax**
- 1 in 3 people with BHD get **kidney cancer**

CUTANEOUS MANIFESTATIONS

Fibrofolliculomas are benign epidermal tumours originating from hair follicles. They appear as white growths on the skin of the head and upper torso. They often develop in early adulthood, and an individual may have none, few or several hundred over their lifetime. There is currently no permanent treatment, though temporary cosmetic treatments are available. Other dermatological symptoms may also include angiofibromas and oral papules.

PULMONARY MANIFESTATIONS

Multiple bilateral pulmonary cysts are common in BHD patients. These cysts are usually asymptomatic and do not generally affect lung function. They are most commonly found at the base of the lungs and are irregular in shape. Due to the propensity for these cysts to rupture, people affected by BHD are at an increased risk of spontaneous pneumothorax. BHD is thought to be the most common cause of familial spontaneous pneumothorax and it has been suggested that screening for an underlying cause in these cases is recommended. There is currently no treatment for BHD-associated cystic lung disease and management is limited to prevention and treatment of pneumothorax.

RENAL MANIFESTATIONS

Renal tumors in BHD are multifocal and often occur bilaterally. Chromophobe renal cell carcinoma (chRCC) and a mixed pattern of chRCC and oncocytoma account for >70% of BHD-associated tumours. BHD renal tumors are usually slow growing and screening should be offered to all BHD patients from the age of 20, with regular, life-long monitoring to facilitate early diagnosis.