BDH is a rare, autosomal dominant, hereditary cancer syndrome due to germline mutations in the folliculin gene (FLCN). The incidence of BHD is about one in 200,000 of the population; however, it is likely underdiagnosed because of low clinical awareness. Folliculin is a protein expressed on keratinocytes in the basal and spinous layer of the epidermis, dermal fibroblasts, nerve cells, lymphocytes, macrophages and mast cells. Extracutaneously, folliculin is expressed on pulmonary type-1 alveolar epithelial cells and distal nephrons. Folliculin is a tumour suppressor protein, important in cell signalling pathways that regulate cell growth. It is proposed that genetic mutation results in loss of a functional protein in cells, resulting in unchecked tumour growth. The pattern of folliculin expression correlates with the clinical features of BHD, including skin papules of varying pathologies, basal lung cysts, spontaneous pneumothorax and renal cell carcinomas.
Proposed diagnostic criteria for BHD include the presence of one major or two minor features. Major features are at least five adult onset fibrofolliculomas or trichodiscomas, with histological confirmation, or a pathogenic \textit{FLCN} mutation. Fibrofolliculomas and trichodiscomas present as multiple small flesh-coloured or white dome-shaped papules on the face, neck and upper trunk. Angiofibromas have also been associated with BHD; however, the presence of multiple angiofibromas is more suggestive of tuberous sclerosis. Minor features include a history of bilateral basal lung cysts, with or without spontaneous pneumothorax, early onset (age < 50 years) renal carcinoma with multifocal or bilateral carcinomas or a mixed chromophobe/oncocytic histology, and a first-degree relative with BHD.

Patients with BHD have a 50-fold increased risk of primary spontaneous pneumothorax development and more than 80% of affected individuals have multiple pulmonary cysts visible on a computed tomography scan, with the less common basal rather than apical predilection. Renal cell carcinomas occur in 27% of affected individuals. A wide range of renal carcinomas have been reported, with the most common pathology being the unusual mixed oncocytic/chromophobe tumour. More than 65% of patients with BHD and renal carcinoma present with multifocal, bilateral or recurrent tumours.

Early recognition of BHD is crucial, given the high risk of renal neoplasm development which, if detected early, is curable. Thus the general practitioner and dermatologist are uniquely placed to consider the diagnosis of BHD when consulted by a patient requesting removal of multiple facial papules. Unexplained spontaneous pneumothorax and basal lung cysts should also raise suspicion of BHD. Referral for genetic testing and multidisciplinary care is essential. Routine renal radiographic screening is recommended for affected individuals > 40 years and, given the likelihood of multiple tumours, nephron-sparing surgery is preferred.

Treatment of skin lesions associated with BHD is elective. In particular, laser ablation of individual lesions may improve the appearance for cosmetic reasons.

\textbf{Competing interests:} No relevant disclosures.

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References are available online at www.mja.com.au.