2.13 Management and follow-up of Birt-Hogg-Dube Syndrome in a rare lung disease clinic

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Background: Birt-Hogg-Dube Syndrome (BHDS) is a rare disorder of the Folliculin-gene involving systems including the lungs, skin and kidneys. Respiratory involvement is characterised by cysts and spontaneous pneumothorax. Other manifestations include renal cell carcinoma (30% risk), and various skin disorders. Through patients in our rare lung disease clinic, we aim to characterise the clinical manifestations and follow-up required for these patients.**Methods:** Patients with BHDS in our Rare Lung Disease clinic were recruited and their charts, management plans and previous investigations reviewed. **Results:** Screening is performed on index-case's families, including genetic testing, pulmonary function testing, HRCT-Chest and MRI-kidneys. Two families were found through this method. One index patient came to our attention following multiple pneumothoraces since age 14, requiring three surgeries. It was then revealed his daughter had a pneumothorax at the age of 14 requiring VATS-pleurectomy. He has 12 identifiable first-degree relatives presently being tested for BHDS. Another family recruited consists of a proband and two brothers undergoing testing.**Discussion:** BHDS is a rare genetic disorder with respiratory, dermatological and oncological/renal involvement. These patients require regular follow-up imaging and family screening. It should be considered in patients with unexplained lung cysts, or families with multiple members suffering from pneumothorax.**Conflict of interests:** Authors declare no conflict of interests