

Birt-Hogg-Dubé Newsletter

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You are receiving this email because you have expressed an interest in BHD. We hope you will enjoy this and future editions. If you do not wish to receive this newsletter, please see the end of the newsletter for instructions.

Safety of air travel in patients with BHD Syndrome

Patients with BHD can have multiple spontaneous pneumothoraces in their life. The lung cysts in BHD can expand in size in a low pressure environment, such as air travel, potentially placing patients with BHD at a higher risk for pneumothorax during air travel. A new study by researchers at the [Rare Lung Disease Consortium](#) aims to investigate the relationship between air travel and the risk of developing a pneumothorax in patients with BHD. This study is questionnaire-based and involves patients with BHD completing an online survey. Further details about the study can be found [here](#).

Getting to know you

This quarter meet Caroline from Germany who was diagnosed with BHD in 2012. The interview can be found [here](#).

BHD Research Highlights

Noteworthy papers from the last quarters include:

Basic:

Starling *et al.* [Folliculin directs the formation of a Rab34-RILP complex to control the nutrient-dependent dynamic distribution of lysosomes](#). EMBO Rep. 2016 Jun;17(6):823-41. [Epub 2016 Apr 13].

- Starling *et al.* show that starvation-induced FLCN association with lysosomes drives the formation of contact sites between lysosomes and Rab34-positive peri-nuclear membranes, by promoting the association of Rab34 with its effector RILP. This restricts lysosome motility and promotes their retention in the peri-nuclear region of the cell.

Iribe *et al.* [Genome-Wide Uniparental Disomy and Copy Number Variations in Renal Cell Carcinomas Associated with Birt-Hogg-Dubé Syndrome](#). Am J Pathol. 2016 Feb; 186(2):337-46

- Iribe *et al.*, assessed chromosomal copy number variation (CNV) and loss of heterogeneity due to uniparental disomy (UPD) in a variety of RCC subtypes from BHD patients. Unlike sporadic tumours the BHD samples showed little or uncharacteristic CNV but high levels of UPD including several common sections potentially identifying a cytogenetic panel for BHD-RCC.

Clinical:

Dong *et al.* [Case Report of Birt-Hogg-Dubé Syndrome: Germline Mutations of FLCN Detected in Patients With Renal Cancer and Thyroid Cancer](#). Medicine (Baltimore). 2016 May; 95(22). [Epub 2016 Jun 3].

- Dong *et al.* report the case of two patients with skin lesions, renal cell carcinoma and papillary thyroid cancer. Genetic analysis was performed and 8 mutations were detected in the *FLCN* gene of the patients. The missense mutations (c.1481A>G and c.1645C>G) were not reported previously. The group suggests that total thyroidectomy may be suitable for thyroid cancer patients with BHD and recommends neck ultrasound for BHD patients and their families.

Kato *et al.* [Fluorescent and Chromogenic in situ Hybridization of CEN17q as a Potent Useful Diagnostic Marker for Birt-Hogg-Dubé Syndrome-associated Chromophobe Renal Cell Carcinomas](#). Human Pathol. 2016 Jun; 52:74-82. [Epub 2016 Feb 4].

- Using fluorescent and chromogenic in situ hybridisation Kato *et al.* determined that the status of chromosome 17q, 2p and 6p can be used to distinguish between sporadic and BHD-associated chromophobe RCC.

Martínez-Pérez *et al.* [Birt-Hogg-Dubé syndrome and colon polyps](#). Rev Gastroenterol Mex. 2016 Jan 29; pii: S0375-0906(16)00002-1 [Article in Spanish]

- Martínez-Pérez *et al.* describe a patients who presented with fibrofolliculomas but no pulmonary or renal pathology. Further testing identified a colonic polyp that was successfully removed surgically.

Review:

Kennedy *et al.*, [Mechanisms of Pulmonary Cyst Pathogenesis in Birt-Hogg-Dube Syndrome: The Stretch Hypothesis](#). Semin Cell Dev Biol. 2016 Apr; 52:47-52. [Epub 2016 Feb 10]

- Kennedy *et al.* review current BHD research relevant to the stretch hypothesis and pulmonary cysts. Reduced FLCN is associated with increased cell-cell adhesion which in areas of enhanced stress could result in cyst formation. Further research is required but advances in recent years are encouraging.

Tellechea *et al.* [Benign follicular tumors](#). An Bras Dermatol. 2015 Dec; 90 (6): 780-98

- Tellechea *et al.*, discuss the development of hair follicles and various benign hair follicle tumour histologies including differential diagnoses. Both trichodiscomas and fibrofolliculomas are discussed as markers of BHD and their importance as potential indicators of a cancer-prone syndrome highlighted.

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