

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.

Rare Disease Day 2017

The [International Rare Disease Day](#) is held on the last day of February every year to raise awareness of rare diseases amongst the public, researchers, healthcare professionals and policymakers. In its tenth-year Rare Disease Day was celebrated in over 80 different countries with events in hundreds of cities. The theme for Rare Disease Day 2017 was *Research* with a campaign inviting people from all over the world to come together to advocate for more research on rare diseases.

New BHD clinical trial on ClinicalTrials.gov

A new BHD clinical trial is currently recruiting patients - [Assessment of Safety of Air Travel in Patients with BHD Syndrome](#). This observation study aims to assess the safety of air travel in patients with BHD via a questionnaire, to further characterise the clinical aspects of disease and to establish a contact registry for these patients to facilitate future studies. Further details about the study are available [here](#).

International Rare Diseases Policy Events

The BHD Foundation attended the Rare Diseases International Policy Event "[The Right to Health: The Rare Disease Perspective](#)" in Geneva and the [3rd International Rare Disease Research Consortium \(IRDiRC\) conference](#) in Paris at the beginning of February to discuss the progress in the rare disease field and to set new goals for the future.

Getting to know you

This quarter meet Filippo from Italy who was diagnosed with BHD in 2009. The interview can be found [here](#).

BHD Research Highlights

Noteworthy papers from the last quarter include:

Basic:

Nagashima *et al.*, 2017. [Nutrient-induced FNIP degradation by SCF \$\beta\$ -TRCP regulates FLCN complex localization and promotes renal cancer progression](#). Oncotarget. 2017 Feb 7.

- Nagashima *et al.* show that degradation of FNIP2 leads to lysosomal dissociation of FLCN and association of mTOR, which promotes the proliferation of renal cancer cells.

Hasumi *et al.*, 2016. [H255Y and K508R missense mutations in tumour suppressor folliculin \(FLCN\) promote kidney cell proliferation](#). 2016 Dec 22.

- Hasumi *et al.* evaluated the FLCN mutations, H255Y and K508R in genetically engineered mice, and demonstrated that they both are pathogenic mutations that promote aberrant kidney cell proliferation to different degrees, with the potential for malignancy.

Clinical:

Sasso *et al.*, 2017. [High resolution computed tomography of the chest in the evaluation of patients with Birt-Hogg-Dubé syndrome](#). Rev Port Pneumol. 2017 Mar 16.

- Sasso *et al.* retrospectively reviewed the clinical records and chest HRCT scans of 9 patients with BHD Syndrome. The authors concluded that pulmonary cysts in imaging studies may be the initial finding in patients with BHD, highlighting the role of CT in early diagnosis.

Gupta *et al.*, 2017. [Spontaneous Pneumothoraces in Patients with Birt-Hogg-Dubé Syndrome](#). Ann Am Thorac Soc. 2017 Mar 1.

- Gupta *et al.* evaluate the risk of pneumothorax, the efficacy of pleurodesis, and the safety of air travel in patients with BHD.

Park *et al.*, 2017. [Birt-Hogg-Dube syndrome prospectively detected by review of chest computed tomography scans](#). PLoS One. 2017 Feb 2.

- Park *et al.* detected BHD Syndrome prospectively in patients undergoing chest computed tomography (CT) scans and classified the characteristics of BHD in Korea.

Li *et al.*, 2017. [Novel germline mutations in FLCN gene identified in two Chinese patients with Birt-Hogg-Dubé syndrome](#). Chin J Cancer. 2017 Jan 9.

- Li *et al.* report two Chinese BHD patients with novel FLCN mutations presenting RCC and spontaneous pneumothorax without fibrofolliculomas and spontaneous pneumothorax on their family history. The incidence of fibrofolliculomas may be lower among Asian BHD patients compared with the higher incidence reported among patients from the US and Europe.

Castellucci *et al.*, 2016. [Multiple chromophobe and clear cell renal cancer in a patient affected by Birt-Hogg-Dubé syndrome: a case report](#). Urologia. 2016 Dec 16.

- Castellucci *et al.* report the case of a patient with seven kidney tumours classified as multifocal type chromophobe renal cell carcinoma and clear cell and spontaneous pneumothorax.

Review:

Jensen *et al.*, 2017. [Birt-Hogg-Dubé syndrome: a case report and a review of the literature](#). Eur Clin Respir J. 2017 Feb 20.

- Jensen *et al.* reviewed the literature on BHD Syndrome on PubMed to give an update on genetics, clinical manifestations, diagnosis, treatment, prognosis and follow-up on BHD.

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