

Birt-Hogg-Dubé Newsletter

September 2015

Vol.14, No.3

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.

Sixth BHD Symposium and First International Upstate Kidney Cancer Symposium 2015

Last week the [Sixth BHD Symposium and First International Upstate Kidney Cancer Symposium](#) took place in Syracuse, New York. Highlights from the Symposium will be available on the website soon. Thank you to Professor Gennady Bratslavsky, Dr Mehdi Mollapour and their team at Upstate for organising such an excellent conference and to all those who participated to make it a success.

Clinical trial of Everolimus in BHD Patients

The [NIH clinical centre](#) in Bethesda has announced a clinical trial to assess the safety and efficacy of everolimus in BHD patients. Participants will receive a daily everolimus tablet for up to a year and have regular check-ups with the trial team. The trial is currently recruiting patients and more details can be found [here](#).

Getting to know you

This quarter meet Maria from Brazil who was diagnosed with BHD in 2015, and Dr Nishant Gupta who is a clinical researcher at the University of Cincinnati. Nishant is a pulmonologist with an interest in BHD and other rare cystic lung diseases, and aims to raise awareness of BHD to increase diagnosis rates in pneumothorax patients. The interviews can be found [here](#).

BHD Research Highlights

Noteworthy papers from the last quarter include:

Basic:

Ding *et al.* [Promoter methylation is not associated with FLCN irregularity in lung cyst lesions of primary spontaneous pneumothorax](#). Mol Med Rep. 2015 Sep 21. [Epub ahead of print]

- The presence of pulmonary cysts in the mid and lower regions of the lung is characteristic of BHD but is also seen in some sporadic pneumothorax patients. Ding *et al.* determined that there is variable expression of FLCN in some of these patients compared to controls. This variation was found not to be associated with mutations in FLCN or altered promoter methylation.

Clinical:

Baken *et al.*, [Birt-Hogg-Dubé syndrome: A diagnosis to consider in patients with renal cancer and pulmonary cysts](#). Diagn Interv Imaging. 2015 Aug 18. [Epub ahead of print].

- Baken *et al.* review a BHD case study identified following thoracic CT and histological analysis of renal tumours. They stress the need for radiologists to be more familiar with BHD enabling early diagnosis in those with characteristic pulmonary cysts.

Ebana *et al.* [Pleural Covering Application for Recurrent Pneumothorax in a Patient with Birt-Hogg-Dubé Syndrome](#). Ann Thorac Cardiovasc Surg. 2015 Sep 11. [Epub ahead of print].

- Ebana *et al.* report the successful treatment of a patient with recurrent pneumothorax using the pleural covering technique. Application of regenerative oxidised cellulose and fibrin glue can help to

thicken and strengthen the lung surface reducing the incidence of pneumothorax without permanently attaching the lung to the chest wall.

Kamada *et al.* [Perifollicular fibromas associated with Birt-Hogg-Dubé syndrome](#). J Dermatol. 2015 Sep 21. [Epub ahead of print].

- Kamada *et al.* describe perifollicular fibromas on the lower back of a Japanese patient, genetically confirmed to have BHD, with a history of recurrent pneumothorax but no other cutaneous manifestations. It has been suggested that perifollicular fibromas are part of the same clinical spectrum as fibrofolliculomas and the authors suppose they could be also be associated with BHD.

Näf *et al.* [Birt-Hogg-Dubé syndrome: novel FLCN frameshift deletion in daughter and father with renal cell carcinomas](#). Fam Cancer. 2015 Sep 5 [Epub ahead of print]

- Näf *et al.* report a novel *FLCN* frameshift and truncating mutation, c.563delT, originally identified in a female with bilateral chromophobe RCC and pulmonary cysts identified by CT. It was also found in her father, with a history of renal cancer, and her two asymptomatic sons who will be monitored for tumour growth.

Tamburrini *et al.*, [Simultaneous Bilateral Spontaneous Pneumothorax Revealed Birt-Hogg-Dubé Syndrome](#). Case Rep Surg. 2015 Sep 1 [Epub ahead of print]

- Tamburrini *et al.* report the first case study of BHD diagnosed in a patient presenting with simultaneous bilateral spontaneous pneumothorax. Characteristic subpleural pulmonary cysts and a family history of pneumothoraces led to a suspicion of BHD, which was confirmed by genetic testing. Identification of other affected family members enabled early detection of RCC in the patient's brother.

Review:

Schmidt & Linehan [Molecular genetics and clinical features of Birt-Hogg-Dubé syndrome](#) Nat Rev Urol. 2015 Sep 1 [Epub ahead of print]

- In this comprehensive review Schmidt and Linehan discuss the clinical manifestations, diagnostic criteria, management strategies and potential therapies in BHD. Additionally they review the current genetic understanding of BHD, the known functions of *FLCN* and important tumourigenic pathways.

To participate in an interview feature, submit information or suggest a topic for the next newsletter, please contact us at contact@BHDSyndrome.org.

To unsubscribe, send an email to contact@bhdsyndrome.org; write "UNSUBSCRIBE" in the subject line of the email.

