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.

Sixth BHD Symposium and First International Upstate Kidney Cancer Symposium 2015

Preparations for the Sixth BHD Symposium and First International Upstate Kidney Cancer Symposium in Syracuse, New York, on 23rd-26th September are ongoing but please be aware that the deadline for registration is today (June 30th). For more information see our website and travel grants are available. We look forward seeing as many of you there as possible.

BHDSyndrome.org updates

To help increase patient and clinician awareness of BHD the patient information pamphlets and "Clinical Introduction to BHD" are now available in French, German, Spanish, Italian, Mandarin and Japanese.

Getting to know you

This quarter, meet Jan from the USA who was diagnosed with BHD in 2013, and Long Yi who is a clinician and researcher in at Nanjing University Medical School. Long Yi is part of the BHD Workgroup in China who are working to increase understanding of the role FLCN mutations play in apparent sporadic pneumothoraces and to increase BHD diagnoses. The interviews can be found here.

BHD Research Highlights

Noteworthy papers from the last quarter include:

Basic :

Chen *et al*., Disruption of tubular Flcn expression as a mouse model for renal tumor induction. Kidney Int. 2015 June 17 [Epub ahead of print]

- Chen *et al*., report on the development of a new mouse model of BHD that recapitulates several features of BHD-RCC. It could be used to increase understanding of tumourigenesis and for screening new drugs to treat BHD-related tumours.

Clinical:

Aivaz *et al*., Comedonal and Cystic Fibrofolliculomas in Birt-Hogg-Dube Syndrome. JAMA Dermatol. 2015 May 13. [Epub ahead of print].

- Aivaz *et al.* suggest that BHD be considered in the differential diagnosis of comedomal and cystic fibrofolliculomas following the identification of such growths in BHD patients. The authors also stress the importance of early diagnosis with regards to future treatment and screening.

Bondavalli *et al.,* Is cardiac rhabdomyoma a feature of Birt Hogg Dubé syndrome? Am J Med Genet A. 2015 April.

- Bondavalli *et al.* report on an interesting BHD case study; an infant who presented with a cardiac rhabdomyoma and was subsequently determined to carry a *FLCN* mutation. Bondavalli *et al*. suggest BHD be included in cardiac rhabdomyoma differential diagnoses but stress that more studies are required to determine a link between BHD and cardiac pathologies.

Ding *et al.,* FLCN intragenic deletions in Chinese familial primary spontaneous pneumothorax. Am J Med Genet A. 2015 May ;167(5):1125-33.

- Ding *et al*. report on the identification of new large deletions in the FLCN genes of families with a history of pneumothoraces. These large deletion were carefully mapped and found to be flanked by Alu elements. Haplotype analysis suggested they are founder mutations in the Chinese population studied.

Kapoor *et al.,* Birt-Hogg-Dubé syndrome and intracranial vascular pathologies. Fam Cancer. 2015 May 8. [Epub ahead of print]

- Kapoor *et al.* report a case series of vascular anomalies in BHD patients. The authors suggest that increased HIF signalling and MMP-9 expression, both associated with vascular malformations and dysfunction, resulting from reduced FLCN could link these pathologies to BHD.

Nishida *et al*., Possible familial case of Birt-Hogg-Dubé syndrome complicated with lung cancer: A possible link between these two disease entities. Respir Med. 2015 May 14. [Epub ahead of print]

- Nishida *et al*. report a case study of a woman with multiple pulmonary cysts, confirmed to carry a FLCN mutation, who developed an adenocarcinoma in one lung. Both she and her brother, who had a history of pneumothoraces and lung cancer, were smokers and the authors suggest a link between FLCN mutations and lung tumours in smokers.

Okada et al., Partial Pleural Covering for Intractable Pneumothorax in Patients with Birt-Hogg-Dubé Syndrome. Clin Respir J. 2015 June 15 [Epub ahead of print]

- Okada *et al*. report on the successful surgical treatment of three BHD patients, with a history of recurrent pneumothorax, using partial lung covering. Following VATS-assisted wedge resection, bioabsorbable PGA felt or ROC mesh were used to cover all excision sites and remaining unruptured cysts. The patients have suffered no recurrent pneumothoraces since surgery (30-32 months).

Review:

Gupta *et al.,* Diffuse Cystic Lung Disease: Part I and Part II Am J Respir Crit Care Med. 2015 Apr 23 [Epub ahead of print]

- In a two part review Gupta *et al*. discuss the pathogenesis, pathology, diagnosis and management of a range of diffuse cystic lung diseases (CLD). BHD is discussed in Part II as the focus of CLD due to genetic mutations.

Ha *et al.,* Cystic lung disease: Systematic, stepwise diagnosis. Cleve Clin J Med. 2015 Feb, 82(2): 115-127.

- Ha *et al.* provide the basis for a step wise diagnosis of cystic lung diseases, including BHD, following computed tomography. The conditions are discussed in terms of clinical presentation and characteristic imaging features.

Richards *et al.,* Cystic and Nodular Lung Disease. Clin Chest Med. 2015 June, 36(2): 299-312.

- Richards *et al.* review the characteristic CT imaging findings that can be used to diagnose a range of CLDs, including BHD, and differentiate them from conditions that mimic CDL. In addition the diagnosis of nodular lung diseases based on history, physical examination and imaging is discussed.

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

BHD foundation