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.

Introducing the BHD Foundation

As the Myrovlytis Trust's work in BHD syndrome has increasingly overlapped with work on related diseases, we have created a new subsidiary to streamline our activities. The BHD Foundation is a registered charity which will oversee all BHD-exclusive projects, allowing the Myrovlytis Trust will broaden its remit to include related rare genetic kidney disorders.

BHD Research Blog survey

The BHD Research bloggers would like to hear your thoughts about the blog. Please take a moment to complete a short survey here. Your participation is very much appreciated, and by way of thanks, you can enter into a draw for £500 towards attendance at the 4th BHD Symposium. The survey closes on the 1st of November 2011.

Connect with the BHD community

BHDSyndrome.org has joined Twitter. Follow our tweets to keep informed about the latest BHD research, BHD Research Blog posts and to get in touch with the wider rare disease community.

Several new interviews filmed at the Third BHD Symposium have been posted online. Hear from a cross-section of leaders in BHD research, from Canada, the United States, the Netherlands and the UK. Keep watching the Video Interviews page as more will be coming soon!

New BHDSyndrome.org resources

Acting on suggestions from the BHD community, these new resources are now on BHDSyndrome.org:

- Updated Information pamphlets to download and print. The Clinical Introduction pamphlet is a useful overview to give to anyone when explaining BHD syndrome. The Diagnosis Information pamphlet offers a guide to obtaining an accurate diagnosis and the implications of a positive genetic test for BHD. The other pamphlets provide information on BHD symptoms and current treatment options.
- New photos of patients with fibrofolliculomas. These have been posted to Skin Symptoms, Fibrofolliculomas, Diagnosis of Skin Symptoms, and Treatment for Skin Lesions. A sincere thank you to Dr Derek Lim and the authors of Menko *et al*. (2009), who kindly provided the photos, as well to the individuals who agreed to share these photos.

New Research Funding

We are delighted to announce several grant awards:

Dr Douglas Medvetz of Brigham and Women's Hospital in Boston, United States, has received a new grant to investigate the role of Folliculin in lung and skin tumour formation.

A new grant has been awarded to Dr Andrew Tee of Cardiff University, UK, to enable identification of possible therapies for BHD.

The Myrovlytis Trust has recently partnered with the Michigan High Throughput Screening Center to undertake a first-stage drug screening project for BHD syndrome. This project will identify classes of compounds which have the potential to be developed as therapeutics and treat BHD syndrome in the future.

Getting to know you

This quarter, meet Dr Andrew Tee, who leads a research group at Cardiff University in Wales, and Helma from the Netherlands, who was diagnosed with BHD syndrome in 2010.

BHD Research Highlights

Noteworthy papers from the last quarter include:

CLINICAL:

Kasi *et al*, 2011. Birt-Hogg-Dubé syndrome: answering questions raised by a case report published in 1962. Case Rep Oncol. 2011 May;4(2):363-6.

- Case study of a patient diagnosed with BHD syndrome after presenting with multiple medical findings consistent with BHD. A study of the patient was first published in 1962 as an unusual case of air embolism brought on by decompression; he was diagnosed with pulmonary cysts at that time. Authors stress that physicians need to have a suspicion of BHD syndrome when they see the clustering of particular medical conditions, especially unexplained spontaneous pneumothorax.

Starink *et al*, 2011. Familial multiple discoid fibromas: A look-alike of Birt-Hogg-Dubé syndrome not linked to the FLCN locus. J Am Acad Dermatol. 2011 Jul 25. [Epub ahead of print]

- Authors describe the novel condition Familial multiple discoid fibromas (FMDF) after studying 9 families with related symptoms. FMDF may have a similar clinical presentation to BHD, but the two conditions are proposed to be distinct. There is no apparent renal or pulmonary involvement in FMDF. The discoid fibromas which characterise FMDF show distinct pathology from BHD-associated fibrofolliculomas. The authors find that FMDF is not linked to the FLCN locus.

Alonso-Gonzàlez *et al,* 2011. Birt-Hogg-Dubé syndrome in a patient with localized fibrofolliculomas and a novel mutation in the FLCN gene. Int J Dermatol. 2011 Aug;50(8):968-71.

- Case report of a 64 year-old woman who presented with skin lesions localised to the right side of the neck. Colonoscopy, abdominal ultrasound and abdominal thoracic scans were performed; findings were limited to renal and hepatic cysts. A novel *FLCN* mutation was identified and the authors suggest that the localisation of fibrofolliculomas represents a mild form of BHD syndrome.

Hopkins *et al,* 2011. Recurrent pneumothorax. Lancet. 2011 May 7;377(9777):1624.

- Authors present a report of a 38 year-old woman with a history of recurrent spontaneous pneumothorax. A *FLCN* mutation was detected and the patient was diagnosed with BHD. The need to consider BHD as a diagnosis for cases of pneumothorax, even in the absence of other characteristic symptoms, was emphasised.

If you would like to participate in an interview feature, please contact us at contact@BHDSyndrome.org
If you would like to submit information or suggest a topic for the next newsletter, please contact the editor at info@BHDsyndrome.org