

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.

Third BHD Symposium, Maastricht, May 2011

The [Third BHD Symposium](#) was a successful and energetic meeting with over 80 participants, including both BHD and HLRCC researchers and patients. Summaries of the [scientific proceedings](#) of the Symposium, as well as of the [patient and family sessions](#), are now available on the [BHD research blog](#). Thank you to all who attended and contributed to the success of the Symposium.

Additionally, a great thank you to all who completed the Symposium survey. Your responses are much appreciated and continue to be helpful as planning proceeds for the Fourth BHD Symposium. The Fourth BHD Symposium will most likely take place in North America in April of 2012.

Forum messaging system

A new messaging system has been developed for the [Forum](#) on BHDSyndrome.org. From next week registered Forum users will be able to send and receive personal messages from one another. Messages will be sent to the email listed in their profile, but email addresses will remain confidential. . This will be an opt-in system - if you would like to receive messages, please let us know by emailing contact@BHDSyndrome.org.

Researcher video interviews

The last newsletter introduced the first video interview on BHDSyndrome.org, with [Professor Eamonn Maher](#). Now [four new interviews](#) have been posted online, with scientists from many aspects of BHD research: [Dr Ravi Nookala](#), [Dr Ferenc Mueller](#), [Dr Richard Harbottle](#) and [Dr Andy Tee](#). In addition, new interviews filmed at the Third BHD Symposium will be available online soon.

If there are any interviews or blog posts you like, please do not hesitate to press the like button at the end of each post!

Information pamphlets

Information pamphlets will also be available soon. Thank you very much to all who have taken the time to complete the [pamphlet survey](#). All your comments have been very useful and constructive. The deadline has been extended until Sunday July 3rd, 11:45 pm British Summer Time, so there is still time to share your suggestions.

New Research Funding

The Myrovlytis Trust is delighted to have awarded a grant extension to Professor Sir Tom Blundell and Dr Ravi Nookala of the University of Cambridge, UK. The Blundell group will continue to extend their work elucidating the structure and function of Folliculin.

The Myrovlytis Trust is also delighted to announce a new grant awarded to Dr Rachel Giles at the University Medical Centre, Utrecht, the Netherlands to study the possible effect of PCT124 on FLCN mutations.

Getting to know you

This quarter, meet [Dr Laura Denby](#), a post-doctoral researcher at the University of Glasgow working on renal gene therapy, and [Anita](#) from the United States, who was diagnosed with BHD syndrome in 2007.

BHD Research Highlights

Noteworthy papers from the last quarter include:

BASIC SCIENCE:

Nahorski *et al*, 2011. [Birt Hogg-Dubé syndrome associated FLCN mutations disrupt protein stability](#). *Human Mutations*. 2011 Apr 28. Epublished ahead of print.

- Analysed the impact of infrequent missense and inframe-deletion *FLCN* mutations on protein function to identify candidate critical functional domains. Performed *in silico* evolutionary analysis of *FLCN* sequence and found that the protein sequence has been highly conserved, particularly the region between codons 100-230.

CLINICAL:

Tobino *et al*, 2011. [Differentiation between Birt-Hogg-Dubé syndrome and lymphangioleiomyomatosis: Quantitative analysis of pulmonary cysts on computed tomography of the chest in 66 females](#). *European Journal of Radiology*. 2011 May 5. Epublished ahead of print.

- Studied CT scans of 14 female patients with BHD and 52 female patients with lymphangioleiomyomatosis (LAM) to determine if features of the pulmonary cysts could be specifically associated with either condition and therefore aid diagnosis. Family history of pneumothorax, location of cysts, diffusing capacity, and size of cysts were proposed as most significant factors in differentiating BHD and LAM.

Boris *et al*, 2011. [The Impact of Germline BHD Mutation on Histological Concordance and Clinical Treatment of Patients With Bilateral Renal Masses and Known Unilateral Oncocytoma](#). *Journal of Urology*. 2011 Apr 13. Epublished ahead of print.

- Investigated the incidence of oncocytoma in one kidney with at least one tumour of same histology in the other kidney by analyzing 23 patients with confirmed BHD and 17 negative for BHD. The 23 BHD patients were found to have significantly lower histological concordance. Authors suggested that BHD patients therefore require more specialised treatment options.

Van Denhove *et al*, 2011. [\[Multiple spontaneous pneumothoraces revealing Birt-Hogg-Dubé syndrome\]](#). *Revue des maladies respiratoires*. 2011 Mar. Article in French.

- Case study of a patient diagnosed with BHD syndrome after presenting with multiples episodes of pneumothorax. The authors strongly suggested that BHD syndrome should be considered in the differential diagnosis of lung cysts.

If you would like to participate in our 'Getting to know you!' 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

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