

Birt-Hogg-Dubé Newsletter

December 2011

Vol.10, No.4

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.

Fourth BHD Symposium

We are pleased to announce that the [Fourth BHD Symposium](#) will be held in Cincinnati, Ohio, USA, on 28th-30th March 2012. The programme will include keynote talks, abstract presentations, and a poster exhibition. Additionally, there will be parallel sessions for patients and families.

The abstract deadline is the 15th February 2012.

Registration and abstract submission will be open soon. Do check the [Fourth BHD symposium page](#) for all updates.

BHDSyndrome.org survey

Let us know if there is anything to be improved for BHDSyndrome.org. There is a new [feedback form](#) that will always be available for you on the [Contact Us](#) page to share your thoughts and suggestions on any aspect of BHDSyndrome.org. [Have a look](#) now.

Additionally, a thank you to all who completed the [BHD Research Blog](#) survey. The winner of the prize draw for contribution towards attendance at the [Fourth BHD Symposium](#) has been contacted. For those who participated in the prize draw, please be sure to check your spam folder. Do continue to follow the [BHD Research Blog](#) to keep up-to-date with what is happening in the BHD field.

Researcher video interviews

Watch the latest interview with [Dr Marston Linehan](#), Chief of the Urologic Oncology Branch at the NIH in the United States. This video is the last in the current series. Visit the [video interviews](#) to see any you may have missed and hear from scientists working on understanding the function of Folliculin, a researcher in gene therapy or a genetic counsellor. There are also transcripts and audio-only files available for each video.

New Research Funding

We are delighted to announce two grant awards:

Professor Maria Czyzyk-Krzeska of the [University of Cincinnati](#) in the United States has received a new grant to investigate the protein networks of Folliculin.

Miss Laura Pradella of the [University of Bologna](#) has been awarded a two-year post-doctoral grant to study the development of oncocytic renal tumours in BHD and Cowden syndrome.

Getting to know you

This quarter, meet [Dr Suet-Ping Wong](#), a post-doctoral researcher working on non-viral gene therapy for BHD, and [Michelle](#) from the United States, who was diagnosed with BHD syndrome in 2006.

BHD Research Highlights

Noteworthy papers from the last quarter include:

CLINICAL:

Houweling *et al*, 2011. [Renal cancer and pneumothorax risk in Birt-Hogg-Dubé syndrome; an analysis of 115 FLCN mutation carriers from 35 BHD families](#). Br J Cancer. 2011 Dec 6;105(12):1912-9.

- This large-scale study reassessed the risk of renal cancer and pneumothorax in BHD. The authors determined a 16% increased lifetime risk of renal cancer and 29% increased lifetime risk of pneumothorax for *FLCN* mutation carriers. Additionally, the majority of renal tumours showed characteristics of eosinophilic variants of clear cell and chromophobe carcinoma.

Truchuelo *et al*, 2011. [Multiple Facial Papules of Birt-Hogg-Dubé Syndrome Treated with a CO\(2\) Laser](#). Ann Dermatol. 2011 Oct;23(Suppl 2):S279-80.

- The authors reported on the use of CO₂ laser treatment for fibrofolliculomas and angiofibromas in two patients clinically diagnosed with BHD. Over 75% of the lesions were removed. One patient was followed-up for 18 months and the other for 4 years; during this time lesions did not recur.

Linguraru *et al*, 2011. [Automated noninvasive classification of renal cancer on multiphase CT](#). Med Phys. 2011 Oct;38(10):5738.

- 125 renal lesions from patients with BHD, VHL, HPRC and HLRCC were tested to study the potential of automated classification of renal lesions from CT. The authors reported that the computer-aided clinical tool which can analyse lesion shape and enhancement allowed accurate quantification of solid, cystic and mixed renal tumours. This tool could be useful as an additional method of non-invasive classification and monitoring of renal tumours.

Sattler *et al*, 2011. [Late Onset of Skin Manifestations in Birt-Hogg-Dubé Syndrome with FLCN Mutation p.W260X](#). Acta Derm Venereol. 2011 Nov 9.

- Sattler *et al*. reported the case of a 48 year-old man diagnosed with BHD who developed fibrofolliculomas at age 40, and was also found to have a kidney lipoma. His 51-year-old brother was also diagnosed with BHD; both were identified to carry the mutation p.W260X. This mutation was previously identified in a Swiss family which exhibited only lung symptoms, with no renal or cutaneous manifestations.

Park *et al*, 2011. [Genetic study in a case of birt-hogg-dubé syndrome](#). Ann Dermatol. 2011 Oct;23(Suppl 2):S188-92.

Shin *et al*, 2011. [Birt-hogg-dubé syndrome, a rare case in Korea confirmed by genetic analysis](#). Ann Dermatol. 2011 Oct;23(Suppl 2):S193-6.

- Park *et al*. and Shin *et al*. presented two cases of BHD in Korea. Park *et al*. discussed a case of a 43-year-old man who presented with trichodiscomas and was found to carry a *FLCN* mutation. Shin *et al*. reported a case of a 54-year-old man presenting with fibrofolliculomas and a history of spontaneous pneumothorax, who was also found to be positive for a *FLCN* mutation.

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