

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.

Coming very soon: Third BHD Symposium, Maastricht, 11th and 12th May, 2011

There is now little over a month left before the [Third BHD Symposium](#). Since the last newsletter, further details have been finalised.

For Patients and Families: there will be a day of patient-centred events, including talks by BHD expert clinicians and a genetic counsellor, round-table discussions and time for getting to know one another. In addition, there will be joint sessions for both patients and researchers.

For Researchers: we are delighted to welcome two more keynote speakers: Dr Patrick Pollard and Dr Ian Tomlinson, both from the University of Oxford. Several dozen abstracts have been submitted and a programme will be available soon.

Given the speakers, the outstanding abstracts and the increasing pace of BHD research, we anticipate a very interesting symposium and hope to see many of you in Maastricht on 11th and 12th May!

New BHDSyndrome.org

The new [BHDSyndrome.org](#) is live! New features include our first [video interview](#) with a BHD researcher and a '[BHD Worldwide](#)' map of BHD families around the world. Already people from Italy, Australia, the United States and the United Kingdom have participated, so be sure to put yourself on the map soon!

All content has been updated, such as the '[What is BHD?](#)' researchers' section which incorporates all the latest research up to 2011 and the [Laboratory Essentials](#) section which provides a comprehensive catalogue of resources for the lab.

We hope the website will be a valuable resource for the BHD community. If you have any suggestions or comments, please email contact@bhdsyndrome.org.

New Research Funding

The Myrovlytis Trust is pleased to award a new two-year grant extension to Professor Arnim Pause of McGill University in Montreal, Canada. Professor Pause will be continuing his work on the functional characterisation of Folliculin and determining its relationships in associated signalling pathways.

The Myrovlytis Trust is also pleased to award prof. dr. Maurice van Steensel, dermatologist at the University Hospital Maastricht in the Netherlands, an additional one-year grant to look further at the cellular function of Folliculin.

Getting to know you

This quarter, meet Professor Arnim Pause, who studies the function of Folliculin, and Jocelyn who was diagnosed with BHD Syndrome in 2008; Arnim lives in Canada and Jocelyn is from the USA. The interviews can be found [here](#).

BHD Research Highlights

Noteworthy papers from the last quarter include:

BASIC SCIENCE:

Benhammou *et al*, 2011. [Identification of intragenic deletions and duplication in the FLCN gene in Birt-Hogg-Dubé syndrome](#). *Genes Chromosomes Cancer*. 2011 Mar 15. Published ahead of print.

- Analysed *FLCN* in 23 individuals from 15 unrelated families with clinically confirmed BHD syndrome but negative DNA sequencing results. Detected several intragenic deletions and one intragenic duplication using real-time quantitative PCR (RQ-PCR), multiplex ligation-dependent probe amplification (MLPA), and array-based comparative genomic hybridization (aCGH). These results expand the techniques used to genetically confirm a BHD diagnosis.

Cash *et al*, 2011. [Loss of the Birt-Hogg-Dubé tumor suppressor results in apoptotic resistance due to aberrant TGFβ-mediated transcription](#). *Oncogene*. 2011 Jan 24. Published ahead of print. (Full text available in the [BHD Article Library](#))

- Presented a novel tumour suppressive mechanism for *FLCN*: regulating TGFβ-dependent transcription and apoptosis.

Lu *et al*, 2011. [Therapeutic targeting the loss of the Birt-Hogg-Dubé suppressor gene](#). *Mol Cancer Ther*. 2011 Jan;10(1):80-9.

- Demonstrated mithramycin as the most effective therapeutic to inhibit tumour growth in renal cell carcinoma, out of 15 candidate compounds selected by the COMPARE algorithm, and evaluated by performance in growth inhibition assay.

CLINICAL:

Tomassetti *et al*, 2011. [Pulmonary features of Birt-Hogg-Dubé syndrome: Cystic lesions and pulmonary histiocytoma](#). *Respir Med*. 2011 Feb 26. Published ahead of print.

- Described BHD-associated lung lesions, noting that these may be indistinguishable from lymphangioliomyomatosis in clinical presentation. Also identified a novel mutation in *FLCN*.

Fontcuberta *et al*, 2011. [Choroidal Melanoma and Lid Fibrofolliculomas in Birt-Hogg-Dubé Syndrome](#). *Ophthalmic Genet*. 2011 Mar 14. Published ahead of print.

- Report of a 63-year-old patient previously diagnosed with BHD syndrome, who presents with fibrofolliculomas on the eyelids and choroidal melanoma. This is the first BHD patient described to develop choroidal melanoma.

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