

# 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.*

## Fourth BHD Symposium

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The [Fourth BHD Symposium](#) will be concluding today. A great thank you to all! During the past two days in Cincinnati, USA, over 80 family members, researchers and health professionals gathered to meet one another and make advances in overcoming BHD. The programme has included keynote talks by Dr Marston Linehan of the [National Cancer Institute](#) at the NIH, Professor Arnim Pause of [McGill University](#), Professor George Thomas of the [University of Cincinnati](#), prof .dr. Maurice van Steensel of the [University Hospital Maastricht](#) and Professor Kuniaki Seyama of [Juntendo University](#).

Continue to follow the Symposium news live on [Twitter](#) with the hashtag #BHDSyndrome. Additionally, see the [BHD Research Blog](#) for an on-site update on the Symposium; more detailed reports summarising the highlights will be posted on the blog soon. We have also been filming more [video interviews](#) at the Symposium, which will be available to view in early summer.

If you attended the Symposium, your thoughts and suggestions are very important for planning the next meeting. Please complete a [feedback survey here](#). The deadline is Monday, the 16<sup>th</sup> of April.

## Information Standard

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The Myrovlytis Trust is pleased to have been awarded certification by the [Information Standard](#), to recognise that it is a provider of high quality, safe and reliable health information. The Information Standard is an independent certification scheme for health and social care information, supported by the [Department of Health](#) in England.

## New MyrovlytisTrust.org

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[MyrovlytisTrust.org](#) has recently undergone an update in look and content. We are also pleased to introduce a new way to play a part in our research programme: you can now choose to [donate](#) to a specific research project. Visit the [new site](#) to learn more. Your feedback is welcome: please email [contact@myrovlytistrust.org](mailto:contact@myrovlytistrust.org).

## New Research Funding

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We are delighted to announce the following grant awards:

Dr Richard Harbottle of [Imperial College London](#) has received a grant for continued support of his project in non-viral renal gene therapy.

Dr Seung-Beom Hong of the [University of Pennsylvania](#) has been awarded a grant continuation to investigate the protein interactions of FLCN.

Professor Stephen Land of the [University of Dundee](#) in the United Kingdom has received a new grant to investigate the pathogenesis of the lung symptoms of BHD.

## Getting to know you

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This quarter, meet [Dr Doug Medvetz](#), who is a post-doctoral research fellow investigating the protein interactions of Folliculin at [Brigham and Women's Hospital](#), Boston, as well as [Drew](#) from the United States, who was diagnosed with BHD syndrome in 2007.

## BHD Research Highlights

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Noteworthy papers from the last quarter include:

### BASIC:

Lim *et al*, 2011. [Activation of AMP-activated protein kinase by MAPO1 and FLCN induces apoptosis triggered by alkylated base mismatch in DNA](#). DNA Repair (Amst). 2011 Dec 29.

- The authors studied the activity of MAPO1 (FNIP2) in *N*-Nitroso-*N*-methylurea-induced apoptosis. Their findings suggest that FNIP2 in complex with FLCN may regulate the activation of AMPK to induce apoptosis.

### CLINICAL:

Barrisford *et al*, 2011. [Familial renal cancer: molecular genetics and surgical management](#). Int J Surg Oncol. 2011;2011:658767.

- The authors reviewed the genetics, molecular pathways, and surgical management guidelines for familial renal cancer syndromes, including von Hippel-Lindau disease, hereditary papillary renal carcinoma, hereditary leiomyomatosis renal cell carcinoma, and BHD.

Furuya *et al*, 2012. [Pulmonary Cysts of Birt-Hogg-Dubé Syndrome: A Clinicopathologic and Immunohistochemical Study of 9 Families](#). Am J Surg Pathol. 2012 Apr;36(4):589-600.

- The authors reported a study of BHD-associated pulmonary cysts in 11 individuals from 9 families. Furuya *et al* found that BHD-associated lung cysts can be distinguished from nonspecific cysts, with features such as internal septa of alveolar walls, an inner lining of epithelial cells and positive staining for phospho-S6 ribosomal protein suggesting mTOR pathway activation.

Renfree and Lawless. 2012. [Multiple Neurilemmomas in Birt-Hogg-Dubé Syndrome: Case Report](#). J Hand Surg Am. 2012 Feb 9.

- Renfree and Lawless reviewed the case of a woman diagnosed with BHD who presented with multiple neurilemmomas in one hand. The authors suggested that hand surgeons consider a diagnosis of BHD in patients with multiple neurilemmomas, particularly in the presentation of associated spontaneous pneumothorax, renal tumours or other neoplasms.

Verhaert LLW. 2011. [A Young Man with Bilateral Spontaneous Pneumothorax](#). Case Reports in Pulmonology. Volume 2011, Article ID 414165.

- Verhaert discussed the case of a young man who presented with bilateral spontaneous pneumothorax and a family history of pneumothorax, as well as pulmonary cysts upon examination. No skin lesions were identified. The young man was found to have a mutation in exon 10 of *FLCN*. The author stressed that BHD should be considered as a diagnosis with patients presenting with spontaneous pneumothorax and a family history of pneumothorax.

**To participate in an interview feature, submit information or suggest a topic for the next newsletter, please contact us at [contact@BHDSyndrome.org](mailto:contact@BHDSyndrome.org).**

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