

Introduction

to the

M Y R O V L Y T I S T R U S T

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Introduction

The Myrovlytis Trust (www.myrovlytistrust.org) is a new medical research charity based in London. Created in late 2007, we aim to:

1. Promote research into rare genetic disorders, including but not limited to Birt-Hogg-Dubé (BHD) syndrome;
2. Advance education of the public in medical and molecular genetics.

We currently focus on BHD syndrome.

What is BHD syndrome?

BHD syndrome is a rare, monogenic (folliculin), autosomal dominant disorder. Symptoms include skin lesions, pulmonary cysts with a risk of pneumothorax, and renal cell carcinoma (RCC). For unknown reasons, symptoms vary between individuals, with RCC occurring in up to 25% of cases. In the UK, about thirty families are known to have BHD. It is believed to be under-diagnosed, but it is unclear by how much. Diagnosis was originally by histological analysis of the skin lesions, but is now typically by DNA sequencing.

What do we do?

1. Promote an international BHD research community. We aim to bring together resources from around the world to facilitate communication, and collaboration, and to shorten the time required to produce results. Accordingly, we have helped to set up a network of European BHD researchers - the European BHD Consortium (EBC) www.EuropeanBHDConsortium.eu - which we hope will:
 - a. Promote greater collaboration within the field of BHD syndrome and greater interaction between BHD researchers;
 - b. Raise the profile of BHD syndrome;
 - c. Make lab resources (antibodies, etc.) publicly available to BHD researchers.

2. Fund basic research. The biology underlying BHD syndrome is unclear, so we are funding basic laboratory research. To date, we have awarded grants worth >£1m.
3. Fund travel grants. Grants are awarded to enable researchers to attend meetings, or to engage in research, related to BHD syndrome. Funds are strictly limited. Further details and the application form are here: www.myrovlytistrust.org/tg.aspx.
4. Enable 'Inaugural BHD Symposium'. This will be held in Roskilde, Denmark, on September 3rd 2008. The Scientific Organising Committee includes the EBC and is led by Professor Eamonn Maher and Dr Laura Schmidt. We are organising the meeting in collaboration with the BHD Family Alliance (an American patient group – www.BirtHoggDube.org), and we are part-funding it. We hope this meeting will be an opportunity for researchers and clinicians to present their work, share ideas, set up research collaborations, and establish EU treatment guidelines for BHD. The meeting is the day before the 8th International Medical Symposium on VHL, at the same location. We chose this location and timing because many people are interested in both conditions and will already be travelling to the VHL meeting. More details and registration here: <http://eventus.tripus.se/bhd2008>
5. Renal Gene Therapy meeting. We are organising and solely funding a small, closed meeting of leading gene therapists and kidney specialists from around the world, in London in August. We hope this meeting will identify current barriers to progress and how to overcome them.

What are our plans?

1. BHD website. In collaboration with the BHD Family Alliance we are developing a website about BHD syndrome – www.BHDSyndrome.org. We hope this will be the first point of reference for anyone interested in BHD syndrome – families, researchers etc.
2. Therapy. We are considering supporting a small clinical trial for one of the symptoms. We are trying to develop a drug assay for folliculin function. Researchers at the NIH have shown that a conditional folliculin knockout mouse line develops enlarged, polycystic kidneys. Encouragingly, rapamycin slows the abnormal kidney growth. Although there are important differences between this mouse model and the human condition, these experiments are certainly encouraging.

We are also considering ideas for rare diseases in general, and are keen to talk to people with an interest in these areas, including:

3. Estimating rare-disease prevalence. BHD is probably under-diagnosed, so are very interested in novel approaches to estimate rare disease prevalence; approaches that would also be applicable to other rare diseases. One of the aims of the EBC is to develop a database of cases of BHD within the EU.

4. Further 'thought-leader' meetings. We hope that the gene therapy meeting will be the first in a series of symposia, each of which would invite leading global experts, in an area of specific interest to the Myrovlytis Trust, to identify current barriers to progress and generate creative, collaborative ways of overcoming them.
5. Biobank / rare disease infrastructure. Should we support current bio/tissue bank initiatives? Additionally, should we support some sort of rare disease infrastructure: perhaps facilitating clinical trials?
6. Contributing to public debate. How best can we support current or new initiatives to ensure that rare diseases have their deserved prominence in public and policy-making agendas?