



LAUNCHING THE FIRST REGISTRY FOR BIRT-HOGG-DUBE SYNDROME



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The BHD Syndrome International Registry (BIRT) is the patient registry for Birt-Hogg-Dubé syndrome (BHD) managed by the Myrovlytis Trust and BHD Foundation and is powered by Pulse Infoframe.

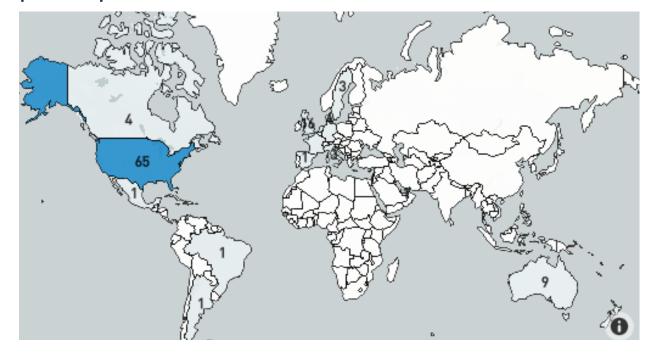
BHD is a rare, inherited condition caused by mutations in the gene folliculin and is characterised by benign skin lesions called fibrofolliculomas, lung cysts and collapsed lungs, and an increased risk of kidney cancer.

BIRT AIMS

- Improve quality of life for BHD patients.
- Facilitate faster diagnoses and reach a consensus on diagnosis and management guidelines.
- Help develop treatments or a cure for BHD.

RESULTS: NUMBERS

As of 30th May 2022, there are 120 active participants across 4 continents.



On average, patients observed their first symptom of BHD at a median age of 30 years old.

However, they did not receive a diagnosis until age 44.

Importantly, over 90% of patients received regular scans of their kidneys. Patients received their first scan shortly after diagnosis (age 44.9).

FUTURE OF BIRT

Year 1 Recruitment of BHD patients

Year 2 Begin work on consensus clinical guidelines Year 3

Year 4 Continue to Expand build data in registry to registry and include data from increase clinicians research

ESTABLISHING BIRT

1. Held patient focus group to identify unmet needs of the community.

2. Formed working group consisting of clinicians and patients.

Received feedback from working group and edited survey questions. 3. Designed medical history survey questions.

5. Finalised survey questions with Pulse Infoframe.

4. Identified and partnered with Pulse Infoframe.

6. Developed registry using the Pulse Platform.

User testing of the Pulse Platform.

7. Launched BHD Syndrome International Registry in March 2022

RESULTS: SYMPTOMS



Genetics

7 out of 10 people saw a genetic counsellor as part of their diagnosis.



Skin

8 out of 10 people had skin lesions.



Kidneys 1 in 6 people had kidney cancer.



Lungs 7 out of 10 people had lung cysts.

TAKE HOME MESSAGES

- Collaboration between advocacy groups, industry, patients and clinicians is necessary and essential in designing and launching a successful registry.
- The large age gap between onset of symptoms and diagnosis represents diagnostic odyssey common to many rare conditions.
- Find out more: myrovlytistrust.org | bhdsyndrome.org | birt.healthie.net | pulseinfoframe.com