

# LAUNCHING THE FIRST REGISTRY FOR BIRT-HOGG-DUBÉ 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.

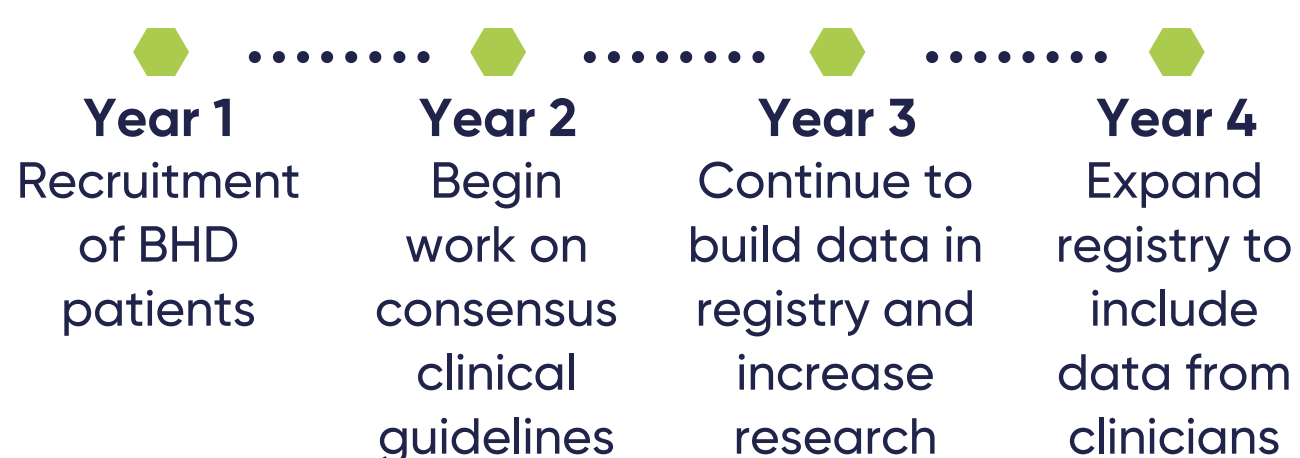


On average, patients observed their first symptom of BHD at a median age of **30** years old.

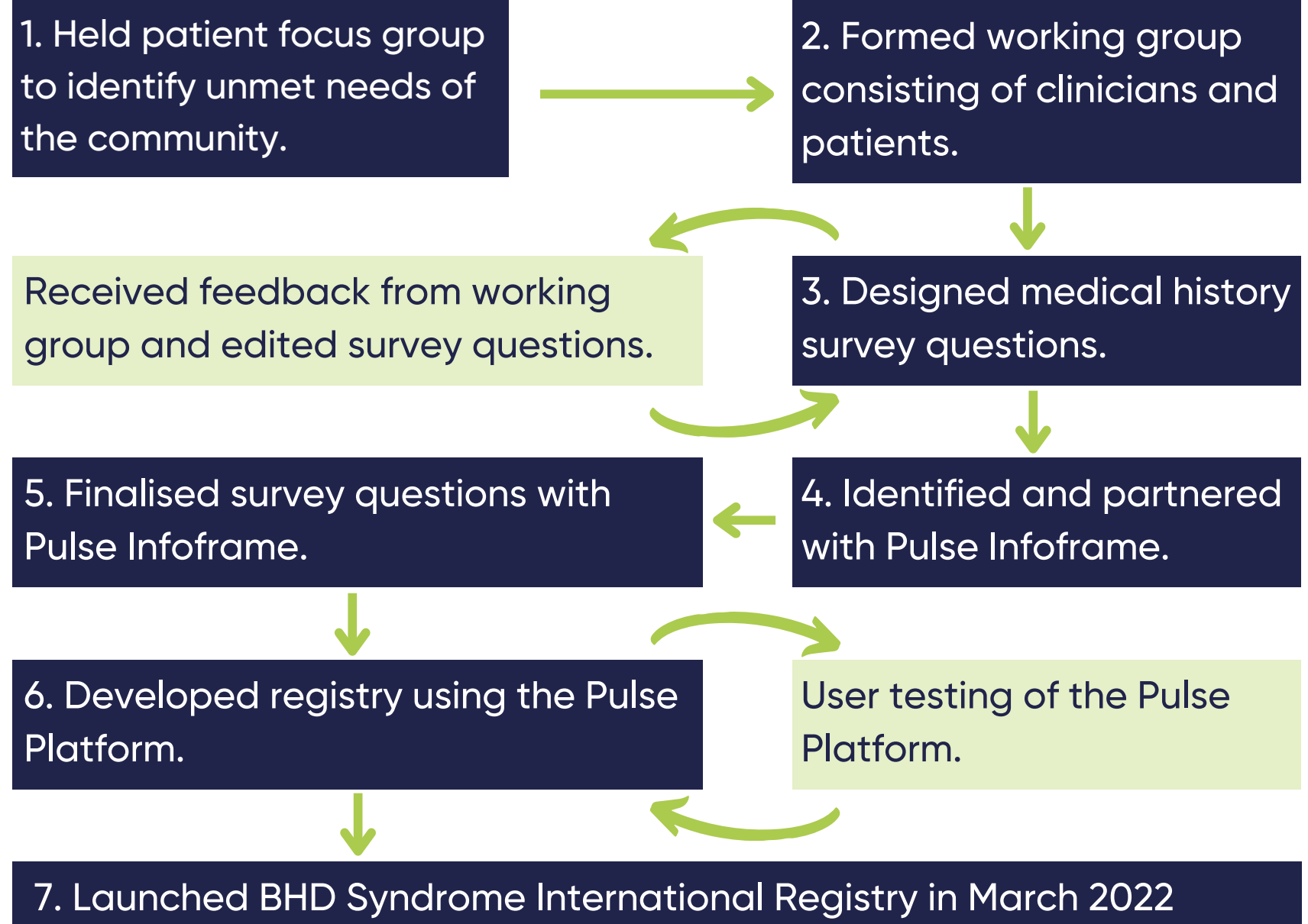
However, they did not receive a diagnosis until age **44**.

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



## ESTABLISHING BIRT



## RESULTS: SYMPTOMS



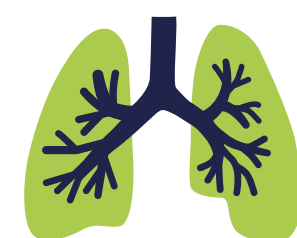
**Genetics**  
**7 out of 10** people saw a genetic counsellor as part of their diagnosis.



**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](http://myrovlytistrust.org) | [bhdsyndrome.org](http://bhdsyndrome.org) | [birt.healthie.net](http://birt.healthie.net) | [pulseinfoframe.com](http://pulseinfoframe.com)