Birt-Hogg-Dubé (BHD) syndrome Care pathway		
*The Patient Clinical Pathway is "the whole care pathway from identification, diagnostics, and multidisciplinary case discussions to surveillance and interventional surgery", so indeed a pathway in time, focusing on **HOW***		
Annual Review Recommended		
BHD syndrome is characterized by an increased risk of renal cell carcinoma, lung cysts and spontaneous pneumothorax as well as cutaneous fibrofolliculomas / trichodiscomas.		
At time of diagnosis, or possible diagnosis, all patients should be offered counselling by a medical genetic specialist. In adulthood, annual (or 2-yearly in some centres) review for renal cell carcinoma should be undertaken by specialists in BHD syndrome (according to national organisation, e.g. radiologists, urologists, geneticists, pulmonologists, and occasionally dermatologists). Surveillance should be continued throughout the person's life or until deemed appropriate based on individual assessment. Assessment and counselling by pulmonologists and dermatologists can be considered.		
Review Checklist — from age 20 years		
	WHAT TO LOOK FOR	**WHEN REFERRAL and WHERE TO**
KIDNEY	Renal tumours	Every 1-2 year and lifelong. Evaluation should be performed by radiologists familiar with BHD syndrome and preferable with MRI with contrast, otherwise ultrasound. In case of abnormal findings or need for treatment refer to urologists familiar with BHD syndrome. No surgical indication for tumours < 3cm. Nephron-sparing surgery whenever possible.
SKIN	Fibrofolliculomas / trichodiscomas	At diagnosis or on patient's request refer to dermatologist, preferably with experience in managing BHD syndrome. Clinicians should consider procedures to remove skin lesions to lessen cosmetic and psychological impact of fibrofolliculomas/trichodiscomas
LUNG	Lung cysts and pneumothorax (collapsed lung)	For activities that may pose a risk for lung collapse expert advice should be sought. These include working as a pilot, flying in unpressurised aircraft or diving. Refer to pulmonologist familiar with BHD syndrome for specialised counselling.
OTHER TUMOURS	Other tumours including familial aggregation of tumours should be handled in accordance to available guidelines for individuals without BHD syndrome.	Refer to relevant specialty.
PSYCHOLOGICAL BURDEN	Psychological issues related to BHD syndrome.	Psychological problems are likely underestimated. Patients may need encouragement to address these issues. Consider referral to psychologist familiar with BHD syndrome or other inherited cancer predisposition syndromes.
PREGNANCY	Familial risk of BHD syndrome.	Pre-implantation genetic testing and/or invasive testing is technically possible. Men and women who are planning pregnancy should be offered referral to a medical genetics specialist for information and (reproductive) counselling.

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Birt-Hogg-Dubé (BHD) syndrome

Care pathway

Family name:

Given name(s)

Address:

Date of Birth: Sex: ☐ M ☐ F ☐ I

Annual Review Recommended

BHD syndrome is characterized by an increased risk of renal cell carcinoma, cutaneous fibrofolliculomas / trichodiscomas as well as lung cysts and spontaneous pneumothorax.

At time of diagnosis, or possible diagnosis, all patients should be offered counselling by a medical genetic specialist.
In adulthood, annual (or 2-yearly in some centres) review for renal cell carcinoma should be undertaken by specialists in BHD syndrome (according to national organisation, e.g. radiologists, urologists, geneticists, pulmonologists, and occasionally dermatologists).
Surveillance should be continued throughout the person's life or until deemed appropriate based on individual assessment.
Assessment and counselling by pulmonologists and dermatologists can be considered.

BHD syndrome Review Checklist

Clinical Presentation:
..... ☐
Other symptoms:
.....
.....
.....
Genetic counselling completed ☐
Date Completed: .....
Clinical diagnosis
.....
Genetic Test '+'ve ☐
Diagnosis Date: .....

General Health Check:
Please record the following information as soon as possible and then annually:

Height
.....

Weight
.....

Blood Pressure
.....

Notes:
.....
.....
.....
.....
.....

Doctor: .....
.....
Review date: .....
.....
Faculty: .....
.....

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WHAT TO LOOK FOR	WHEN TO REFER
KIDNEY: Renal cysts and tumours.	Every 1-2 years from age 20 years and lifelong. Evaluation should be performed by radiologists familiar with BHD syndrome and preferable with MRI with contrast, otherwise ultrasound. In case of abnormal findings refer to urologists familiar with BHD syndrome. No surgical indication for tumours < 3cm. Nephron-sparing surgery whenever possible. ☐ **Date Referred**: .....
SKIN: Fibrofolluculomas / trichodiscomas	At diagnosis or on patient's request refer to dermatologist familiar with BHD syndrome. ☐ **Date Referred**: .....
LUNG: Specialised counselling.	For activities that may pose a risk for lung collapse expert advice should be sought. These include working as a pilot, flying in unpressurised aircraft or diving. Refer to pulmonologist familiar with BHD syndrome for specialised counselling. ☐ **Date Referred**: .....
OTHER TUMOURS: Other tumours including familial aggregation of tumours should be handled in accordance to available guidelines for individuals without BHD syndrome.	Refer to relevant specialty. ☐ **Date Referred**: .....
PREGNANCY: Invasive testing and/or pre-implantation genetic testing is technically available.	When planning pregnancy men and women should be offered referral to a medical genetics specialist. ☐ **Date Referred**: .....
PSYCHOLOGICAL BURDEN: Psychological problems are likely underestimated. Patients may need encouragement to address these issues.	According to individual assessment consider referral to psychologist familiar with BHD syndrome or other inherited cancer predisposition syndromes. ☐ **Date Referred**: .....

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