Protocol for diagnosis, genetic testing and surveillance in individuals with Birt-Hogg-Dubé syndrome

This guideline for the diagnosis, surveillance and management of people with Birt-Hogg-Dubé syndrome has been drawn from the best available evidence and the consensus of experts in this area and it is regularly updated to reflect changes in evidence.

The expectation is that clinicians will follow this guideline unless there is a compelling clinical reason to undertake different management, specific to an individual patient.

European Reference Network
for rare or low prevalence complex diseases

Network
Genetic Tumour Risk Syndromes (ERN GENTURIS)

www.genturis.eu

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The diagnosis of BHD syndrome should be considered in	Strength*	Recommendation
• Primary spontaneous pneumothorax	Strong	1a
• Multiple pulmonary cysts.	Strong	1b
• Bilateral or multifocal renal neoplasia.	Strong	1c
• Renal cell carcinoma, below age 50 or familial.	Strong	1d
• Multiple cutaneous papules consistent with fibrofolliculomas/trichodiscomas	Strong	1e
• Any combination of the above mentioned manifestations in an individual or in the family.	Strong	1f
Genetic testing for BHD syndrome should be offered in		
• Primary spontaneous pneumothorax, if recurrent or familial.	Strong	6a
• Multiple pulmonary cysts in the absence of a known cause.	Strong	6b
• Bilateral or multifocal renal neoplasia.	Strong	6c
• Early onset (usually defined as <45 years) or familial renal cell carcinoma.	Strong	6d
• Multiple cutaneous papules consistent with fibrofolliculomas/trichodiscomas and at least one histologically confirmed.	Strong	6e
• Any combination of the above mentioned manifestations in an individual or in the family with or without a known family history of BHD syndrome.	Strong	6f

Surveillance protocol	Exam	Age	Interval		
• Renal cell carcinoma	Renal MRI	20 y and life-long	Every 1-2 years	Strong	11, 13, 13a, 13b, 14
• Fibrofolliculomas/ trichodiscomas	Consideration of the need for a dermatologic evaluation	At diagnosis	When needed	Strong	18

* This grading is based on published articles and expert consensus: strong – expert consensus AND consistent evidence, moderate – expert consensus WITH inconsistent evidence AND/OR new evidence likely to support the recommendation, weak – expert majority decision WITHOUT consistent evidence.