

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.



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

The diagnosis of BHD syndrome should be considered in				Strength *	Recommendation
<ul style="list-style-type: none"> • Primary spontaneous pneumothorax • Multiple pulmonary cysts. • Bilateral or multifocal renal neoplasia. • Renal cell carcinoma, below age 50 or familial. • Multiple cutaneous papules consistent with fibrofolliculomas/trichodiscomas • Any combination of the above mentioned manifestations in an individual or in the family. 				Strong	1a
				Strong	1b
				Strong	1c
				Strong	1d
				Strong	1e
				Strong	1f
Genetic testing for BHD syndrome should be offered in					
<ul style="list-style-type: none"> • Primary spontaneous pneumothorax, if recurrent or familial. • Multiple pulmonary cysts in the absence of a known cause. • Bilateral or multifocal renal neoplasia. • Early onset (usually defined as <45 years) or familial renal cell carcinoma. • Multiple cutaneous papules consistent with fibrofolliculomas/trichodiscomas and at least one histologically confirmed. • 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	6a
				Strong	6b
				Strong	6c
				Strong	6d
				Strong	6e
				Strong	6f
Surveillance protocol	Exam	Age	Interval		
<ul style="list-style-type: none"> • Renal cell carcinoma 	Renal MRI	20 y and life-long	Every 1-2 years	Strong	11, 13, 13a, 13b, 14
<ul style="list-style-type: none"> • 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.