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ERN GENTURIS PLAIN LANGUAGE SUMMARY OF CLINICAL PRACTICE GUIDELINES FOR THE DIAGNOSIS, SURVEILLANCE AND MANAGEMENT OF PEOPLE WITH BIRT-HOGG-DUBÉ SYNDROME

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Disclaimer: The content of this plain language summary is based on the "ERN GENTURIS CLINICAL PRACTICE GUIDELINES FOR THE DIAGNOSIS, SURVEILLANCE AND MANAGEMENT OF PEOPLE WITH BIRT-HOGG-DUBÉ SYNDROME_public_v1 ".

INTRODUCTION

Individuals with Birt-Hogg-Dubé (BHD) syndrome may develop lung cysts, lung collapses, facial skin bumps and/or kidney tumours. So far there has been no universally agreed consensus on how BHD syndrome should be managed, and thus clinical care for people with BHD syndrome may vary between centres. Guidelines can help people with, and suspected of having, BHD syndrome access appropriate care**.**

GUIDELINE AIMS

The BHD syndrome guideline has been created to assist healthcare professionals to give the most up-to-date recommendations for the diagnosis, management and surveillance of people with BHD syndrome. This guideline has been drawn from the best available evidence, the consensus of experts who care for people with BHD syndrome, and input from people with BHD syndrome. It will be updated to reflect changes in evidence.

SCOPE AND PURPOSE

The guideline is intended to provide the optimal diagnosis, clinical management and surveillance of people with BHD syndrome.

GUIDELINE SUMMARY

The diagnosis of BHD syndrome should be considered in the case of:
• a pneumothorax (lung collapse) with no clear cause. • multiple cysts in the lungs with no known cause. • multiple tumours in one or both of the kidneys. • kidney cancer before the age of 50 or kidney cancer that runs in the family. • facial or upper torso skin bumps diagnosed as fibrofolliculomas/ trichodiscomas • any combination of the above symptoms in the same person or members of their family.
Genetic testing for BHD syndrome should be offered in the case of:
• repeated pneumothoraces (lung collapses) with no clear cause. • a family history of lung collapses with no clear cause. • multiple lung cysts with no known cause. • multiple tumours in one or both of the kidneys. • early onset or a family history of kidney cancer. • skin bumps with at least one confirmed as a fibrofolliculoma/trichodiscomas through skin biopsy testing. • any combination of the above signs or symptoms in the same individual or members of their family.
Surveillance for people with BHD syndrome and *FLCN* mutation carriers should include:

• regular screening for kidney tumours
  ○ from age 20 years and life-long.
  ○ every 1-2 year.
  ○ preferably by MRI with contrast, otherwise with ultrasound.

KEY RECOMMENDATIONS

Kidney	Everyone with BHD syndrome is at risk of kidney tumours which, if not detected and treated can develop into cancer. Everyone should be offered kidney screening. Screening for kidney cancer should: • start at age 20. • be lifelong. • be done every 1 to 2 years using MRI preferably with contrast. Ultrasound can be used if MRI is not available or appropriate. Surgery should usually be done when the largest kidney tumour is 3 cm. Nephron-sparing surgery should ideally be performed whenever possible. This is where only the tumour and a small part of the normal surrounding kidney is removed. Other treatments may be considered where appropriate.
Lungs	Flights on commercial airlines are generally safe for people with BHD syndrome. For activities that may pose a risk for pneumothorax (lung collapse) expert advice should be sought. These include working as a pilot, flying in unpressurised aircraft or diving. Surgical treatments should be considered for the treatment of recurrent pneumothoraces (lung collapses).
Skin	An expert skin examination should be considered for people newly diagnosed with BHD syndrome. For some patients fibrofolliculomas/trichodiscomas affect quality of life. If a patient requests treatment for their fibrofolliculoma/trichodiscoma doctors should consider their options for treatment which may require a referral to a dermatologist.
Other Cancers	Currently there is no strong evidence that BHD syndrome causes any other cancers.

PSYCHOLOGICAL NEEDS

It is important to consider the impact of BHD syndrome on both mental and social wellbeing. Delayed diagnosis, uncertainty about future health problems and/or fear of getting cancer may cause anxiety or depression. Living with a long-term health condition may also have some social challenges. There may be financial concerns such as cost of insurance. The skin features may also cause people to feel self-conscious about the way they look. BHD syndrome might also impact on family relationships. There may be feelings of guilt and concerns when making plans for starting a family.

Addressing the psychological needs of patients and families with BHD syndrome should form a key element of their care. Doctors should ask about wellbeing at each clinical contact and be aware of signs of anxiety and depression. Patients should be reffered for professional support if needed. Peer-to-peer support through patient support groups can also play a key role in wellbeing.