Diagnostic and Interventional Imaging (2019) **100**, 657—658

EDITORIAL

Diagnostic and interventional radiology is a milestone in the management of renal tumors in Birt-Hugg-Dubé syndrome

Initially described in 1977, Birt-Hogg-Dubé (BHD) syndrome is currently defined as an autosomal dominant condition caused by germline mutations in the *FLCN* (folliculin) gene located on chromosome 17p11.2 [1]. BHD syndrome is characterized by skin fibrofolliculomas, multiple lung cysts, spontaneous pneumothorax, and renal tumors. Thus far about 200 families with BHD syndrome with pathogenic *FLCN* mutations have been reported worldwide but BHD syndrome is probably under-diagnosed because of the wide variability in its clinical expression.

A seven-times increase in the risk of renal cancer have been reported in BHD syndrome-affected individuals. Renal tumors are often multifocal and bilateral in more than half of patients but rarely metastatic. Renal cancers occur in up to 30% of the patients during follow-up, at a mean age of 50 years (range 25—75 years). Renal tumors are more frequently chromophobe renal cancer and a mixed pattern of chromophobe and oncocytic renal tumors, while other histological subtypes such as clear-cell and papillary carcinoma, and several mixed patterns have been also reported.

To date, no guidelines have been established far for the clinical management of patients with BHD syndrome [2]. Because of the increased risk of renal cancer, surveillance is indicated for carriers of *FLCN* germline mutations, and their relatives, after 20-year-old. A yearly MRI examination seems the best available surveillance method to detect and characterize renal tumors although ultrasound and computed tomography examinations may be used alternatively [3,4]. Although size, growth rate and location of the tumor(s) will directly affect the therapeutic decision, MRI provide insights of the tumors which will help to avoid biopsies once validated by prospective studies [5,6]. Researchers have demonstrated the potential of multiparametric MRI to characterize noninvasively different renal tumor subtypes, such as chromophobe and oncocytic tumors, the two main tumor subtype encountered in BHD syndrome [7]. By combining all the imaging MR features successively reported in the literature and following practical algorithms based on a step-by-step reading of the MR images, readers are now able to identify several imaging profiles, which appear specific of each renal tumor subtypes and their aggressiveness [8].

https://doi.org/10.1016/j.diii.2019.06.004

As in any other hereditary syndromes such as Von Hippel-Lindau disease with multifocal, bilateral, asynchronous renal tumors, treatment aims to spare renal function and prevent metastatic disease [9]. As demonstrated in the study of Matsui et al. [10], minimally invasive nephron-sparing techniques such as percutaneous radiofrequency ablation and cryoablation might be safely and accurately considered in multidisciplinary team meeting in addition to partial nephrectomy. Relative indications of nephron-sparing surgery and ablations should be discussed based on disease characteristics: size, number and location of the tumor(s) within the kidney as well as patient surgical history and renal function. Based on the growing body of literature on BHD syndrome, surveillance methods, and treatments, studies are now required to establish recommendations avoiding the risk of overtreatment in this population. Diagnostic and interventional radiologists have a unique opportunity to guide the management decisions in BHD syndrome, allowing appropriate screening and counselling of the patients and their relatives.

Disclosure of interest

The author declares that he has no competing interest.

References

[1] Menko FH, van Steensel MA, Giraud S, Friis-Hansen L, Richard S, Ungari S, et al. Birt-Hogg-Dubé syndrome: diagnosis and management. Lancet Oncol 2009;10:1199—206.
[2] Finelli A, Ismaila N, Bro B, Durack J, Eggener S, Evans A, et al. Management of small renal masses: American Society of Clinical Oncology Clinical Practice Guideline. J Clin Oncol 2017;35:668—80.
[3] Lee E, Sayyouh M, Haggerty JE, Kazerooni E, Agarwal PP. Role of radiologists in the diagnosis of unsuspected Birt-Hogg-Dubé syndrome in a tertiary clinical practice. AJR Am J Roentgenol 2019, http://dx.doi.org/10.2214/AJR.19.21176.
[4] Gupta S, Kang HC, Ganeshan D, Morani A, Gautam R, Choyke PL, et al. The ABCs of BHD: an in-depth review of Birt-Hogg-Dubé syndrome. AJR Am J Roentgenol 2017;209:1291—6.
[5] Schieda N, Lim RS, McInnes MDF, Thomassin I, Renard-Penna R, Tavolaro S, et al. Characterization of small (< 4 cm) solid renal masses by computed tomography and magnetic resonance imaging: current evidence and further development. Diagn Interv Imaging 2018;99:443—55.
[6] Cornelis F, Tricaud E, Lasserre AS, Petitpierre F, Bernhard JC, Le Bras Y, et al. Routinely performed multiparametric magnetic resonance imaging helps to differentiate common subtypes of renal tumours. Eur Radiol 2014;24:1068—80.
[7] Galmiche C, Bernhard JC, Yacoub M, Ravaud A, Grenier N, Cornelis F. Is multiparametric MRI useful for differentiating oncocytomas from chromophobe renal cell carcinomas? Am J Roentgenol 2016;208:1—8.
[8] Cornelis F, Grenier N. Multiparametric magnetic resonance imaging of solid renal tumors: a practical algorithm. Semin Ultrasound CT MRI 2016;38:47—58.
[9] Marcelin C, Ambrosetti D, Bernhard JC, Roy C, Grenier N, Cornelis FH. Percutaneous image-guided biopsies of small renal tumors: current practice and perspectives. Diagn Interv Imaging 2017;98:589—99.
[10] Matsui Y, Hiraki T, Gobara H, Iguchi T, Tomita K, Uka M, et al. Percutaneous ablation therapy for renal cell carcinoma associated with Birt—Hogg—Dubé syndrome. Diagn Interv Imaging 2019, http://dx.doi.org/10.1016/j.diii.2019.06.009.

F.H. Cornelis[a,*], J.-C. Bernhard[b]
[a] *Department of Radiology, Sorbonne Université, Tenon Hospital, 4, rue de la Chine, 75020 Paris, France*
[b] *Department of Urology, Bordeaux University, Pellegrin Hospital, place Amélie-Raba-Léon, 33076 Bordeaux, France*
* Corresponding author.
E-mail address: francois.cornelis@aphp.fr (F.H. Cornelis)