J Ped Surg Case Reports 3 (2015) 481–484

Contents lists available at ScienceDirect

Journal of Pediatric Surgery CASE REPORTS

journal homepage: www.jpscasereports.com

Renal oncocytosis in a pediatric patient: Case report and review of the literature

Stacey Speer [a], Daniele Wiseman [a], Madeleine Moussa [b], Andreana Bütter [c,*]

[a] Department of Medical Imaging, London Health Sciences Centre, Western University, London, Ontario, Canada
[b] Department of Pathology, London Health Sciences Centre, Western University, London, Ontario, Canada
[c] Division of Pediatric Surgery, London Health Sciences Centre, Western University, London, Ontario, Canada

ARTICLE INFO

Article history:
Received 5 August 2015
Received in revised form
11 September 2015
Accepted 14 September 2015

Key words:
Renal oncocytoma
Oncocytosis
Pediatric

ABSTRACT

Renal oncocytosis is a rare condition in which the kidney develops numerous oncocytomas. We present a case of a 12-year-old female who presented with right-sided flank pain of one-year duration. Imaging revealed several masses in the right kidney. Tissue biopsy confirmed multiple benign oncocytomas. Due to the presence of multiple oncocytomas throughout the kidney, a radical nephrectomy was performed. Given the rarity of this condition, as well as its known association with von Hippel-Lindau disease and Birt-Hogg-Dube syndrome, genetic investigations were pursued but failed to identify any abnormalities. This patient remains well and disease free six years after surgery. A review of the literature of this rare condition was performed.

Renal oncocytosis is a rare pathological condition in which a wide spectrum of oncocytic changes occur in the kidney [1]. Warfel and Eble initially described oncocytomatosis in an adult patient in whom both kidneys had more than 200 oncocytomas [2]. Tickoo et al. described 14 cases of renal oncocytosis in which each diseased kidney contained numerous oncocytic nodules as well as a dominant mass. This mass is usually a renal oncocytoma although rarely, it can be a chromophobe renal cell carcinoma. Therefore, radical nephrectomy is often warranted due to concern about possible malignancy [3].

Renal oncocytomas are epithelial tumors composed of oncocytes that are well differentiated, contain eosinophilic cells and are arranged in a tubular pattern [1]. Specific genetic conditions, such as Birt-Hogg-Dube (BHD) and Von Hippel-Lindau syndrome, can also be associated with renal oncocytosis [1]. However, very few pediatric cases of renal oncocytoma have been described [4–8].

We report a case of a 12-year-old girl who was found to have multiple renal oncocytomas, bilateral macular degeneration and bilateral sensorineural hearing loss, prompting a thorough genetic investigation. To our knowledge, this is only the 5th reported pediatric patient with renal oncocytosis.

1. Case report

A 12-year-old female presented to her primary care physician with right-sided flank pain of one-year duration. Medical history was significant for bilateral macular degeneration as well as bilateral sensorineural hearing loss. Physical examination was unremarkable. Laboratory investigations revealed a normal complete blood count, liver and kidney function tests. Urinalysis was normal. A nuclear medicine bone scan was unremarkable. Abdominal ultrasound, computed tomography (CT) and magnetic resonance imaging (MRI) were performed. Ultrasound and CT confirmed multiple well-defined complex cystic and solid masses scattered throughout the right kidney (Figs. 1 and 2). The left kidney was normal. The largest mass was located anteriorly within the lower pole measuring 3.7 cm × 4.6 cm (Fig. 1c). This along with several other renal masses showed complex cysts with solid irregular septations. The solid components confirmed flow within them on Doppler suggesting a neoplasm as opposed to a hemorrhagic cyst. Ultrasound imaging confirmed the presence of echogenic shadowing punctate components consistent with calcifications in one lesion. Fat suppressed image sequences on MRI and ultrasound imaging confirmed the lack of macroscopic fat within the lesions thus excluding the diagnosis of angiomyolipomas. No lesion exhibited a classic stellate central scar. In this clinical context and in conjunction with multiple indeterminate unilateral kidney masses in a child, a variety of differential diagnoses were considered

* Corresponding author. Western University, Children's Hospital, 800 Commissioners Rd East, Rm B1-188, London, ON N6A4G5, Canada. Tel.: +1 519 685 8401; fax: +1 519 685 8421.
E-mail address: Andreana.butter@lhsc.on.ca (A. Bütter).

http://dx.doi.org/10.1016/j.epsc.2015.09.009

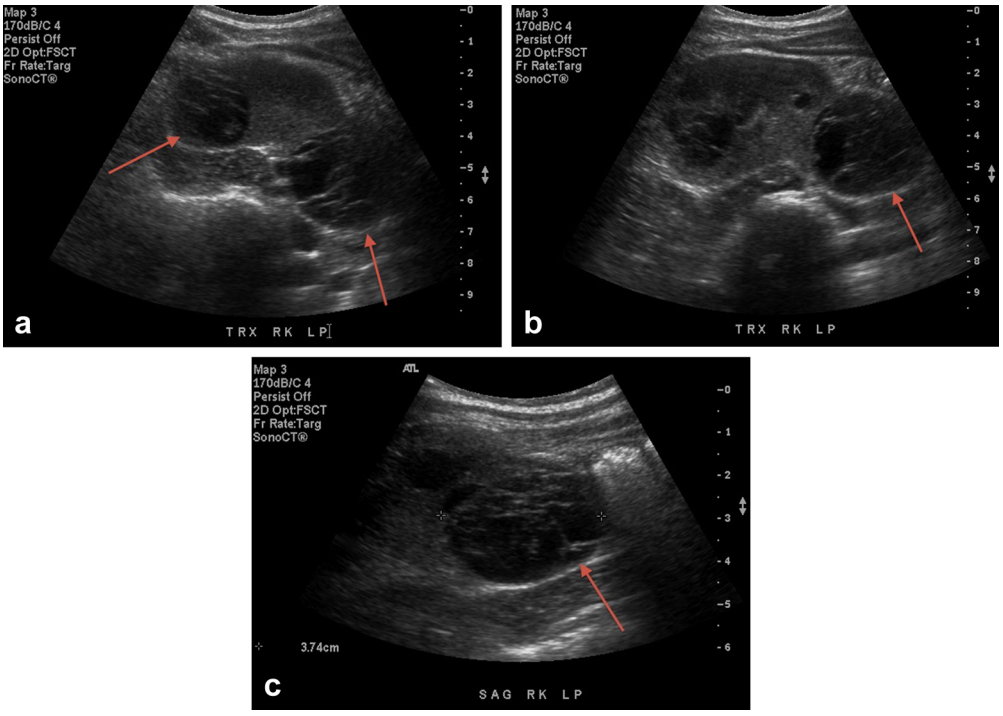

Fig. 1. (a–c) Transverse image of the lower pole of the right kidney demonstrates multiple complex masses. These appear as complex cysts with multiple irregular solid septations in which flow could be seen on color Doppler (not included). The largest was located within the lower pole measuring 3.7 × 4.6 cm.

including: (a) Birt-Hogge-Dube syndrome with renal tumors composed of chromophobe renal cell carcinoma and/or renal oncocytomas, (b) Von Hippel-Lindau disease in the context of multiple renal lesions, (c) tuberous sclerosis with renal angiomyolipomas despite a lack of family history for this diagnosis and lack of macroscopic fat, (d) benign renal oncocytomas and finally (e) renal cell carcinoma.

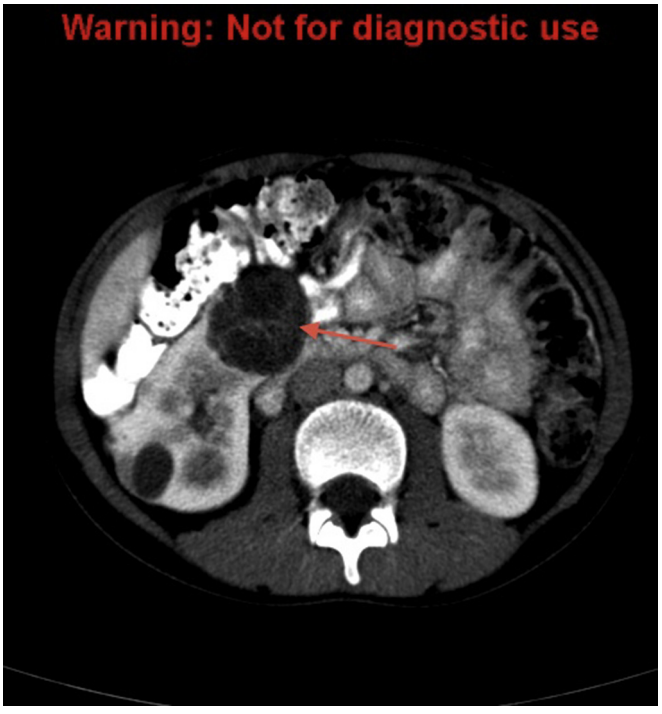

Fig. 2. CT confirms multiple well-defined, low attenuating cystic and solid masses within the right kidney. There is an impression of fine septations configuring into a stellate pattern centrally within the largest lesion but not a classic central scar. There is no fat within these lesions.

Given the diagnostic uncertainty and concern about potential malignancy, the patient underwent laparotomy and open biopsies of her right kidney. Intraoperatively, multiple cystic nodules were noted throughout the kidney. Two biopsies were taken of the largest mass. Frozen section suggested that the lesion was benign (Fig. 3). Definitive treatment was deferred, pending permanent sections. Final pathology revealed neoplastic lesions composed of oncocytic cells arranged in small nests and a tubular architecture with areas of cystic change. The cells appeared monomorphic with minimal nuclear atypia. Mitotic activity was not identified. Clear cytoplasmic change was not identified and there was no necrosis or lymphovascular invasion. The tumor cells were negative for colloidal iron. Immunohistochemistry revealed that the tumor cells stained diffusely for vimentin and epithelial membrane antigen. Entrapped non-neoplastic tubules were positive for cytokeratin 7, while the tumor cells were negative. Rare cells showed cytoplasmic positivity for CD57. Chromographin, synaptophysin, WT-1 stains were negative. Final pathologic diagnosis was multiple right renal oncocytomas and oncocytosis. As a result of these pathologic findings, the diffuse involvement of her entire right renal parenchyma, multiple case reports describing renal oncocytosis as a possible precursor lesion to renal cell carcinoma and extensive discussion with the patient and her mother, a right radical nephrectomy was performed.

Due to the rarity of renal oncocytomas in the pediatric population and its known association with certain genetic conditions (i.e. Birt-Hogg-Dubé and Von Hippel-Lindau syndromes), our genetics team was consulted. In addition, her co-existing bilateral macular degeneration and bilateral sensorineural hearing impairment suggested an underlying genetic abnormality. Chromosomal analysis revealed a 46XX karyotype without any chromosomal abnormalities. Point mutations were not identified on the FLCN gene