Case report and literature review of a rare diagnosis of ossifying renal tumour of infancy

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Abstract

One must entertain a broad differential diagnosis for infants presenting with gross hematuria. Initial workup includes urine analysis, serum laboratory values and abdominal ultrasound. We describe an infant presenting with gross hematuria found to have a calcified renal mass upon initial ultrasound and subsequent computed tomography scan. We considered a differential diagnosis of, but not exclusive to, staghorn calculi, nephroblastoma, Wilms’ tumour, mesoblastic nephroma and ossifying renal tumour of infancy (ORTI). A nephrectomy was performed, and the pathology report identified the calcified mass as an ORTI.

Introduction

There are a wide variety of renal neoplasms in the pediatric population. The differential diagnosis of a renal neoplasm ranges from more common entities, including nephroblastoma, nephroma and Wilms’ tumour, to less frequent ones, such as renal cell carcinoma, and rare entities, such as ossifying renal tumour of infancy (ORTI). It is important to differentiate among different types of renal masses clinically, radiologically and pathologically as their treatment and follow-up differ.

ORTI is a rare, but likely benign, renal tumour, which was first described by Chatten and colleagues in 1980. To date, there are 16 reported cases. We describe a 4-month old boy with ORTI who was treated with a radical nephrectomy. At his 2-month follow-up, he was without recurrence.

Case report

We describe a case of a 4-month old boy who presented with painless gross intermittent gross hematuria since the age of 3 weeks. He had no signs of a urinary tract infection in either clinical or laboratory parameters. Urinalysis demonstrated 100+ red blood cells in high-powered field, and 1+ protein; there was no evidence of a glomerular process. Renal function tests were well within normal limits and no electrolyte abnormalities were present. Abdominal ultrasound was initially negative; however, upon a serial follow-up it revealed a partially calcified appearing mass in the upper pole of the left kidney with no distortion of the renal contour (Fig. 1, part A). There was associated mild pelvicalyceal dilatation. No abnormalities were identified in the other kidney. Doppler examination showed a solitary central signal within the mass. Plain radiograph revealed flecks of dystrophic calcifications in the expected area of the left kidney in the retroperitoneal space. The likely diagnosis of ORTI was based on clinical background and sonographic features. Further characterization of the lesion was achieved by non-contrast and contrast enhanced computed tomography (CT). A CT revealed a 2-cm benign looking partially ossified renal mass within the upper pole of the left kidney with surrounding non-enhancing low attenuation area that was likely a soft tissue component of the mass (Fig. 1, parts B, C and D).

Urologic evaluation was performed by 2 surgeons, with additional input from a pediatric oncologist. A partial nephrectomy was considered; however, given the spiculations extending from the mass on CT, it was felt that enucleation and clear margins may not have been technically feasible. Thus, we decided to proceed with an open subcostal radical nephrectomy. Intraoperatively, no obvious gross extension of the tumour was identified and the procedure was conducted without complication. The patient had an uncomplicated hospital stay and was discharged on postoperative day 3. Pathology results returned with a diagnosis of ORTI, with the mass measuring 2.5 × 2.1 cm (Fig. 2).

At the 3- and 9-month follow-ups, the patient was clinically well with no recurrence of hematuria. He recovered...
from surgery. Repeat abdominal ultrasound was normal with no evidence of recurrence in the ipsilateral or contralateral sides.

**Discussion**

ORTI is a very rare, benign renal tumour. To date, there are 16 reported cases. Although rare, ORTI has certain epidemiological and radiological characteristics that may assist clinicians and radiologists in the diagnosis. The disease typically occurs during infancy, with patient age ranging from 6 days to 30 months. Of the 16 cases of ORTI, most were with boys (75%, n = 12). About 96% (n = 15) of the patients presented with painless intermittent macroscopic hematuria. Although less frequent, an abdominal mass may also be present. Initial imaging investigations typically include ultrasound and CT. An ultrasound typically shows an echogenic mass with shadowing, while CT often shows a well-defined mass with calcification and poor enhancement. Most (75%, n = 12) of the reported cases involved the left kidney. 69% (n = 11) involved the upper pole calyces, and 81% (n = 13) had calcification. The masses are rarely greater than 2 to 3 cm in size. Renal outline is typically maintained, and a filling defect with a local caliectasis is often seen. As our patient exhibited all

**Fig. 1.** Selected ultrasound image of the left kidney with a linear probe (A) demonstrates a well-circumscribed mass in the upper pole (solid white arrow), with internal high echogenicity with associated shadowing (dotted white arrow), representing the pathologic proven ossification within the tumour. Sagittal reformatted computed tomographic (CT) image (B), and axial CT image on soft tissue (C) and bone (D) windows again show the left upper pole mass (solid white arrow), with internal ossification (black asterisk).

**Fig. 2.** Gross specimen, coronal section demonstrating a well-circumscribed 2.5-cm lesion to the left upper pole. Surrounding parenchyma is unremarkable.

**Fig. 3.** Low power image demonstrates osteoblasts producing osteoid on the left. The blastemal cells and native kidney can be seen on the right.
of the above-described demographics, clinical and radiological features, the provisional diagnosis of ORTI was made preoperatively with a high degree of confidence.

Ossification, as seen with ORTI, may easily be mistaken for renal staghorn calculi. However, this diagnosis is exceedingly rare in infancy and thus alternative diagnoses should always be considered. Beyond the infancy period, making the diagnosis of ORTI can become more difficult because having a renal calculus is far more common than having ORTI. Nevertheless, ORTI presents with painless hematuria, and renal calculi are unlikely to be painless.

Other diagnoses on the differential for a calcified renal mass in this particular age group include Wilms’ tumour, nephrogenic rest, renal cell carcinoma, extra-adrenal neuroblastoma, calcified hematoma and post-infectious (e.g., tuberculosis) or post-traumatic changes. Paraneoplastic syndromes, such as hypertension and hypercalcemia, could also be present in some of these disease entities.

Histopathological diagnosis requires the presence of ossification of tissue. Typically, analysis demonstrates an osteoid core, layers of osteoblasts and an epithelial component or spindle cells within the osteoid core. Mature osteoid elements tend to be more prominent in older patients. Despite clear visual characteristics, histogenesis is not well-established for this unusual neoplasm. The origin of the tumour remains unclear, but some believe it arises from the urothelium. Ossification can be the result of the osteogenic potential of the urothelial cells. The ossification and mesenchymal components are similar to mesoblastic nephroma histologically. The mass is described as congruent with the renal medulla, in particular the papillae of the renal pyramids. From this location, it extends into the collecting system in a polypoidal manner. A competing theory suggests that spindle cells resemble intralobar nephrogenic rests and that these lesions may lie within the pathologic spectrum of Wilms’ tumours. However, to date, Wilms’ tumour has not been reported in any of these patients.

No known associations with this tumour have been identified. However, Seixas-Mikelus and colleagues described a 3-month-old male infant with gross hematuria and ORTI in addition to juvenile granulosa cell tumour of the testis. This is the only case report of these 2 lesions presenting concomitantly. The case likely represents a chance finding of 2 distinct disease processes. A potentially newly discovered association cannot be made based on a single case; it requires a more scientific analysis of the disease processes. El-Husseini and colleagues reported a 10-month-old girl with ORTI and asthma. However, this was likely an incidental finding and not related to ORTI.

Although it is considered benign, the true natural history is still a mystery as all reported cases have been treated with surgery, with partial or total nephrectomy. However, Stefeens and colleagues have reported that complete enucleation of the mass without nephrectomy or heminephrectomy may be an alternative treatment strategy. In addition, the use of chemotherapy is not justified, as patients who were only treated surgically had no evidence of recurrence or metastases. The longest follow-up reported was by Ito and colleagues, who had a follow-up of 23 years with no recurrence. Thus, it is important to correctly diagnose ORTI to avoid any unnecessary treatment. However, since only 16 cases have been described, no absolute conclusions may be drawn. Thus, patients should always be followed clinically and with postoperative imaging, such as ultrasound.

Conclusion

ORTI is a very rare renal entity that has some typical clinical, radiological and pathological features. Sixteen reported cases are not significant enough to draw any specific conclusions. However, the current data suggest that a male infant with painless intermittent macroscopic hematuria, with radiological features of a non-invasive calcified renal mass within the upper pole calyceal system, very likely has an ORTI. This constellation of clinical presentation and radiologic findings strongly supports the diagnosis of ORTI prior to pathologic confirmation.

Competing interests: Dr. Flannigan, Dr. Heran, Dr. Oviedo, Dr. Wong and Dr. Masterson all declare no competing financial or personal interests.

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In this study, the authors report a case of ossifying renal tumour in infancy. The patient presented with a palpable abdominal mass, and imaging studies revealed a mass with calcifications. A biopsy was performed, and the histopathological examination confirmed the diagnosis of ossifying renal tumour. The authors reviewed the literature and presented the details of this rare paediatric renal tumour.

References


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