Unusual presentation of more common disease/injury

Bilateral botryoid nephroblastoma: a rare cause of renal failure

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Summary

The authors describe the case of a 16-month-old girl who presented with bilateral hydronephrosis and renal failure secondary to bilateral renal pelvic botryoid Wilms' tumour (nephroblastoma). The term 'botryoid' describes an intrapelvic polypoid renal Wilms tumour, either extending from the renal parenchyma or primarily pelvis-based tumour. Both tumours filled the renal pelvis and extended down the proximal ureter, with no intraparenchymal tumour seen. Bilateral intralobar nephrogenic rests were present. Histological examination demonstrated triphasic nephroblastoma, with focal rhabdomyomatous differentiation of the stromal element bilaterally. Postchemotherapy, the patient underwent bilateral nephrectomy. Post complete resection of her bilateral disease, this patient has had an excellent outcome. This is only the third reported case of bilateral botryoid Wilms' tumour. While the condition is extremely rare, it should be included in the differential diagnosis of a young child with bilateral renal pelvic masses.

BACKGROUND

This is only the third reported case of bilateral botryoid Wilms' tumour (nephroblastoma). While the condition is extremely rare, it should be included in the differential diagnosis of a young child with bilateral renal pelvic masses.

This rare condition presents unusual clinical management challenges, in terms of preservation of residual native renal function, which will be of use to clinicians in similar situations.

CASE PRESENTATION

A 16-month-old girl was admitted to the nephrology department due to renal failure associated with bilateral renal masses. She had presented 2 weeks earlier to her local hospital with a 2 day history of intermittent fever, vomiting, sore throat and coryzal symptoms. Physical examination at that time demonstrated hypertension (138/89) in an irritable, pale child with palpable, bilaterally-enlarged kidneys.

INVESTIGATIONS

Investigations showed microscopic haematuria, renal failure (urea 29.6 mmol/l, creatinine 297 µmol/l, potassium 6.9 mmol/l) and anaemia (haemoglobin 7.9 g/dl).

A renal ultrasound showed bilateral renal pelvic masses, obstructing the pelvi-ureteric junction (PUJ) on both sides, with secondary hydronephrosis.

Initially, a diagnosis of bilateral fungal balls had been considered, but her renal function failed to respond to a trial of antifungal therapy and serial renal ultrasound scans showed no reduction in the size of the masses or the degree of obstruction.

A left antegrade pyelogram, performed during insertion of bilateral percutaneous nephrostomy tubes, demonstrated multiple filling defects in the renal pelvis with no drainage beyond the PUJ.

A presurgical dimercaptosuccinic acid (DMSA) scan revealed a split function of 53% (right) and 47% (left) with a non-functioning left lower pole. No evidence of metastatic disease was identified on CT of chest and abdomen (figure 1).

Bilateral J-J stents were inserted in order to relieve the obstruction, and biopsies were taken.

Histological examination of the bilateral biopsies demonstrated triphasic Wilms' tumour. Background interstitial nephritis was noted, likely secondary to non-steroidal anti-inflammatory drugs (ibuprofen) that she had received for fever before presentation.

TREATMENT

Nephron-sparing surgery was considered for the right sided tumour, but multi-disciplinary review concluded that it was inappropriate due to the central tumour position within the kidney. A decision to proceed with right nephrectomy was made, with intraoperative biopsy of the left kidney. Following an 8 week preoperative chemotherapy course (1 mg/m² vincristine and 30 µg/kg actinomycin D), she underwent right nephrectomy. Biopsies from the left kidney at this operation showed viable nephroblastomatous tissue consistent with a Wilms' tumour. This correlated with the presurgical renal ultrasound which showed only a minor reduction in the size of the left tumour mass postchemotherapy.

During further follow-up, the patient's renal function continued to deteriorate and a repeat renal ultrasound showed an increase in size of the left kidney tumour with extension along the ureter, necessitating left nephrectomy.

OUTCOME AND FOLLOW-UP

Gross inspection of the right nephrectomy demonstrated distension of the ureter by tumour, extending up to 4 cm distal to the PUJ (figure 2). The kidney was distorted by a discrete central mass which filled the renal sinus and pelvis.

Figure 1 Axial image of the bilateral renal pelvic tumours.

Figure 2 Macroscopic image of the botryoid tumour extending down the ureter.

There was no evidence, either macroscopically or histologically, of renal parenchymal involvement. Histological examination demonstrated a triphasic tumour, composed of blastemal, epithelial (tubular structures) and stromal elements, with heterologous rhabdomyomatous differentiation focally (figure 3). Postchemotherapy necrosis

Figure 3 Nephroblastoma demonstrating mesenchymal, epithelial and blastemal elements (white arrows, left to right), and a suburothelial cambium layer (yellow arrow) (H&E).

was present, but treatment response was evident in less than 50% of the tumour. No anaplasia was seen. Multiple intralobar nephrogenic rests were noted adjacent to the tumour, arising in the suburothelial space and demonstrating intrapelvic growth. Genetic analysis confirmed the presence of a constitutional deleterious mutation WT1_c.1168C>T_R390X.

The left nephrectomy demonstrated a tumour which filled and expanded the pelvi-calyceal system and extended down the ureter in a polypoid fashion. Similar to the contralateral tumour, it was a triphasic neoplasm, with focal rhabdomyomatous features and no evidence of anaplasia. Again, intralobar rests were present in the kidney.

The patient had an uneventful postoperative course and commenced on haemodialysis, while continuing with chemotherapy for a further 6 months. The patient has now been in remission for 18 months, and a kidney transplant from a living related donor is planned.

DISCUSSION

Wilms' tumour (nephroblastoma) is one of the most common paediatric solid tumours. The majority of cases are sporadic in nature, although up to 10% are associated with a defined syndrome such as WAGR syndrome (Wilms-aniridia-genital anomaly-retardation syndrome).¹ It derives from nephrogenic blastemal cells, and may develop in pre-existing nephrogenic rests, which can be perilobar or intralobar in position. The vast majority of Wilms' tumours are unilateral, unicentric and located within the renal parenchyma, with no evidence of renal pelvis extension. Such tumours tend to present with a palpable abdominal mass, pain or hypertension. Bilateral intrarenal nephroblastomas are estimated to occur in approximately 5% of cases; bilateral tumour development is associated with younger age at presentation and an increased association with genetic syndromes.

The term 'botryoid Wilms' tumour' has been applied to a number of case reports describing either the extension of an intraparenchymal tumour into the renal collecting

system, or a Wilms' tumour which has arisen within the renal pelvis with no evidence of intraparenchymal extension. In a review of ten reports, Honda *et al*² described four cases of exclusively intrapelvic tumour, without an intraparenchymal component. This phenomenon is rare; to date, 26 individual cases of botryoid nephroblastoma have been the subject of case reports,³ while the National Wilms' Tumour Study Group (NWTSG)⁴ have produced a review of 45 children (enrolled in NWTSG 3, 4 and 5) who were identified as having ureteral tumour extension. In the NWTSG series, the overall incidence of ureteral extension in NWTSG-5 was 2%. Bilateral botryoid nephroblastomas are extremely rare; only two cases have been reported previously.^{3 5} In the first reported case,³ the presenting complaint was haematuria rather than renal failure, and bilateral fibroepithelial polyps (FEPs) were reported in association with the Wilms' tumours. However, since the FEPs are reported to contain 'primitive tubular and glomerular structures' and 'nephroblastomatoid tissues' and they were resected after chemotherapy had commenced, it appears likely that this tissue represented nephroblastoma showing chemotherapy-effect.

Botryoid Wilms' tumours tend to present with haematuria; less commonly, an abdominal mass, fever or painful micturition constitute the presenting complaint. In the NWTSG series,⁴ ureteral extension was detected radiologically before surgery in only 14 of 45 patients; early detection was more common in tumours extending into the lower ureter and bladder. All patients underwent nephrectomy and partial or complete ureterectomy; survival was excellent with complete resection, and did not necessitate postoperative radio- or chemo-therapy.

In this report, we describe an extremely rare form of bilateral botryoid nephroblastoma presenting with acute renal failure associated with fever, hypertension and bilateral palpable abdominal masses. Alternative diagnoses

such as fungal infection were considered initially, due to the rarity of the condition. Various imaging modalities contributed to multi-disciplinary debate before surgical intervention and were vital in the assessment of the feasibility of conservation of some native renal function. The initial hope was to conserve left kidney function, but as the tumour continued to grow despite chemotherapy, left nephrectomy was undertaken and the patient commenced dialysis.

Learning points

- ▶ Wilms' tumour (nephroblastoma), a relatively common paediatric malignancy, may present in unusual ways.
- ▶ Bilateral botryoid Wilms' tumour should be included in the differential diagnosis of a young child with bilateral renal pelvic masses.
- ▶ While every effort should be made to preserve some native renal function in paediatric patients, these tumours may necessitate bilateral nephrectomy.

Competing interests None.

Patient consent Obtained.

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