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CASE REPORT

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Primary lumbosacral Wilms tumour associated with occult spinal dysraphism

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Abstract A 4-year-old child presenting with sudden-onset paraplegia and a sacral tumour in association with spina bifida occulta is reported. There were no stigmata of spinal dysraphism at birth. Imaging studies confirmed a sacral tumour with extradural extension up to T10 and spinal dysraphism. The histological features of the extradural and sacral components of the tumour were consistent with a Wilms tumour. The differential diagnosis included a primary sacral teratoma containing Wilms tumour elements or a primary extrarenal Wilms tumour arising in association with a spinal dysraphism. There was no clinical response to chemotherapy or radiotherapy.

Key words Extrarenal nephroblastoma · Sacrum · Spinal dysraphism · Teratoma · Wilms tumour

Introduction

Spinal malformations due to developmental disturbances occurring at different stages may be separated into dysraphic and nondysraphic malformations [5]. Dysraphic malformations may be further divided into occult spinal dysraphism (spina bifida occulta) or dysraphisms with spina bifida aperta. The occult spinal dysraphisms include the following lesions: long spinal cord, spinal lipoma, diastematomyelia, meningocele, open central canal in the spinal cord or syrinx, neurenteric cyst, dermoid cyst, dermal sinus and tethered cord syndrome [6, 15]. The most common neoplasm seen in association with occult spinal dysraphisms is a teratoma. However, there are few reports of extrarenal Wilms tumour and primitive renal rests occurring in this setting (Table 1). Primary lumbosacral Wilms tumours may arise from heterotopic renal rests of meso- or metanephric origin, neural crest cells, and germ cells or in association with spinal dysraphisms. We report the clinical and histological features of a unique extrarenal lumbosacral Wilms tumour associated with occult spina bifida, which occurred in a 4-year-old child. In addition, we review the literature on heterotopic renal tissue, especially that occurring in association with spinal dysraphisms.

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Clinical history

A 4-year-old girl presented with a 2-day history of sudden onset of paraplegia and total incontinence. Three months prior to this the mother had noticed a sacral mass, which fluctuated in size. At birth, no sacral mass was observed and there were no clinical stigmata of spinal dysraphism. Imaging studies were not performed at birth. The child did not have any neurological symptoms until the onset of paraplegia. There was no other significant history. The child was referred to the neurosurgical unit.

Examination revealed a 6×6 cm area of swelling over the upper end of the natal cleft. She had flaccid paralysis of the lower limbs with a sensory level at T12, decreased anal sphincter tone and no bladder sensation. The upper limbs were normal. The rest of the systemic examination contributed nothing to the diagnosis. A full blood count and electrolytes were normal. Mantoux test, syphilis serology and HIV serology were negative. Chest and thoracolumbar spinal radiographs were reported as normal. Magnetic resonance imaging (MRI) of the spine revealed a long level extradural mixed intensity mass from T10 onwards with extensive paraspinal extension via neuroforamina (Fig. 1). A subsequent computerised tomography (CT) scan revealed a posterior midline soft tissue mass extending from the pelvic brim to the femoral heads, with attachment to the adjacent gluteal muscles. There was also an associated bone defect of the sacrum, which was consistent with a spina bifida occulta (Fig. 2). Further classification of the occult spinal dysraphism was not possible in this case because of extensive intraspinal tumour. No renal masses were detected on ultrasound, CT and MRI investigation.

Table 1 Review of cases of spinal dysraphisms containing renal tissues

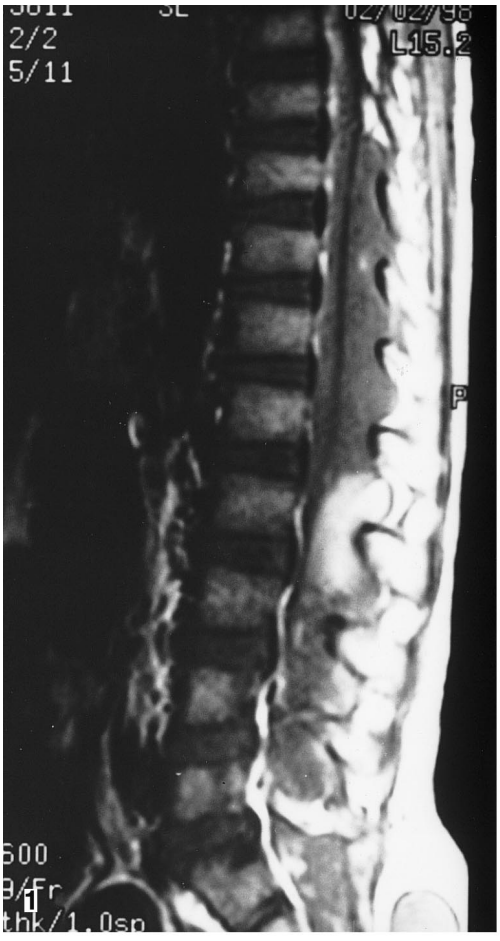
Reference	Age	Gender	Site / dysraphism	Neurological signs	Macroscopic	Renal elements	Treatment	Outcome
[4]	2 years	Female	T12 to L3 / diastematomyelia	Abnormal gait; normal sensation, power and tendon reflexes	3-cm intra-arachnoid mass	Wilms tumour	Surgical excision, chemo- and radiotherapy	Disease free 12 months later
[7]	Neonatal	Female	Diastematomyelia	Not known	Abnormal tissue between paravertebral fascia and dural tube	Immature glomeruli and tubules	Surgical excision	Not known
[8]	2 years	Male	L4 to sacrum spina bifida	Incontinence, leg weakness and hip dislocation	Protruding cystic structure at L4: lipomeningocoele	Immature renal tissue: nephrogenic rests	Surgical excision	Not known
[10]	11 years	Female	L4 to S1 spina bifida	Intact	6×8 cm lumbosacral mass	Wilms tumour	Surgical excision and chemotherapy	Lung metastases disappeared with chemotherapy; patient well
[13]	2 years	Female	T12 to L4 diastematomyelia	Abnormal gait	2.5-cm spherical nodule	Wilms tumour	Surgical excision, chemo- and radiotherapy	Cerebellar metastasis excised; chemo- and radio-therapy. Recurrence free 20 months later
[14]	7 weeks	Female	L5 meningomyelocele	Intact	8-mm disc-shaped lesion	Immature renal tissue – blastema, tubules and glomeruloid structures	Surgical excision	Not known
Present case	4 years	Female	Sacral occult spina bifida	Sudden-onset paraplegia and total incontinence	6×6 cm dorsal sacral mass with extradural extension to T10	Wilms tumour	Nonresectable: chemotherapy and radiotherapy	No response to chemotherapy and radiotherapy. Palliative care

Open spinal biopsy was performed via a L4 hemilaminectomy, and extradural tumour tissue was sent for histological examination. After a month the child was referred for oncological management. Subsequently, the sacral mass was also biopsied. On the basis of the clinical impression of a sacral teratoma, the patient was started on a course of chemotherapy consisting of cisplatin, ifosphamide and etoposide, but there was no neurological improvement. Furthermore, no decrease, in fact an increase, in the size of the mass was noted on physical examination and imaging studies. A course of palliative radiotherapy was commenced, but there was no clinical improvement. The patient was transferred to the referring hospital for palliative care.

Pathological findings

The first specimen of the extradural tumour measured 2×2×3 cm. Microscopic examination showed the characteristic tissue components of a triphasic Wilms tumour. There was a predominance of epithelial structures set in spindle cell mesenchymal tissue. The epithelial structures consisted mainly of primitive tubular structures lined by cuboidal to columnar cells with pleomorphic, hyperchromatic nuclei and scant amounts of cytoplasm (Fig. 3). The tubular cells showed high mitotic activity, with 20 mitoses per 10 high-power fields in some areas. Glomeruloid structures were also present (Fig. 3). In addition, papillary structures growing into small cystic spaces were seen. Small islands of primitive blastema were present focally (Fig. 4). Tumour necrosis was limited to small areas. Anaplasia and other heterologous tissues were absent. There were no rosettes and no neuropil (neurofibrillary matrix), which ruled against neuroblastoma as a differential diagnosis. The histological features were consistent with either a metastatic or a primary extrarenal Wilms tumour. Another possibility was that of a sacral teratoma containing nephroblastomatous tissues, but this seemed unlikely in view of the absence of teratomatous tissue elements.

The above findings together with the absence of a renal mass on imaging studies prompted a second biopsy, this time of the dorsal sacral mass. This biopsy showed the same histological features as the previous biopsy of the extradural tumour. Again there were no heterologous



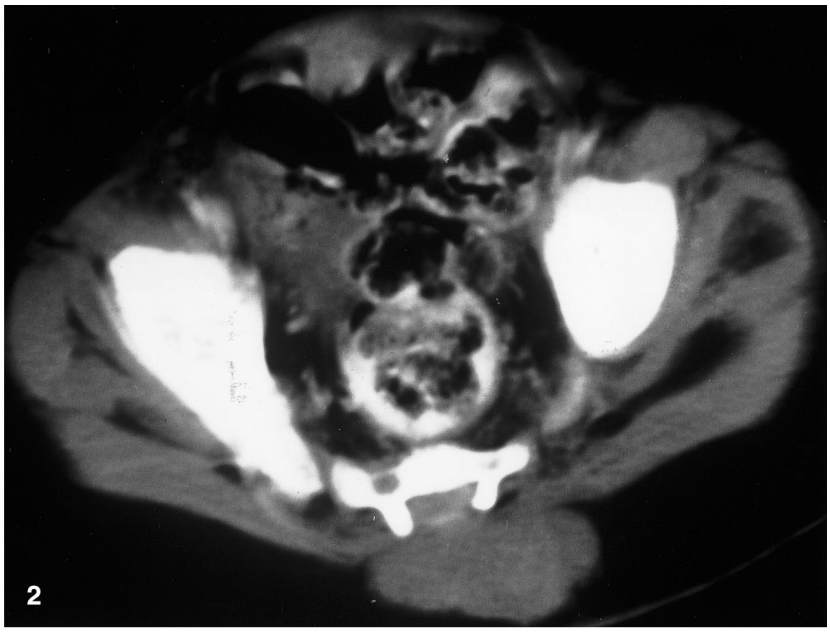
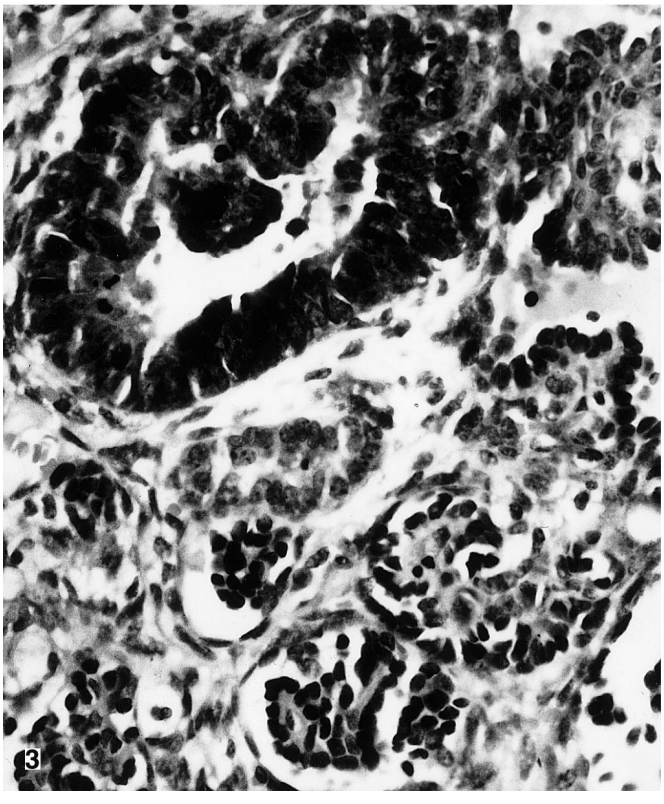


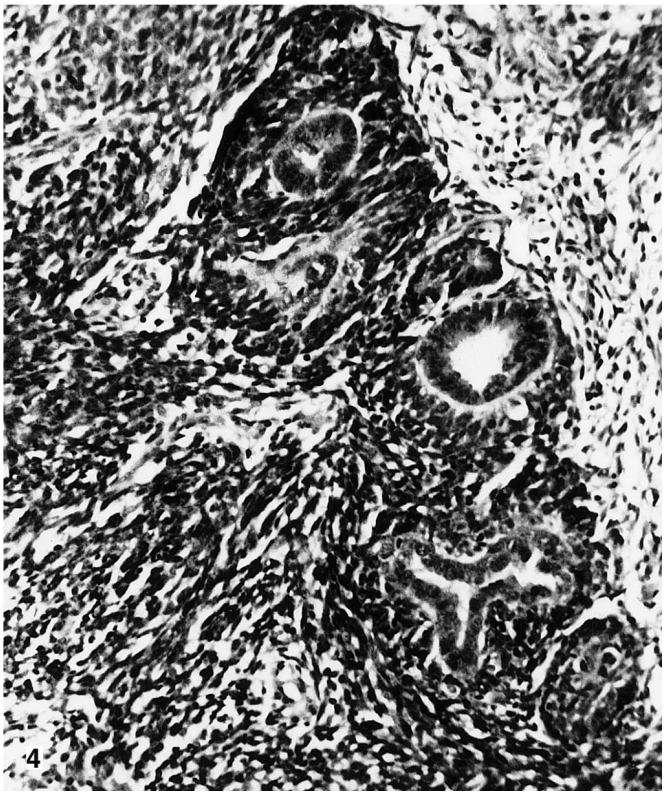
Fig. 1 MRI of the spine showing an extradural mixed intensity mass from T10 onwards

Fig. 2 CT scan showing a bone defect of the sacrum consistent with an occult spina bifida. Note the dorsal sacral mass overlying the bone defect

Fig. 3 Primitive tubular structure lined by columnar cells with pleomorphic, hyperchromatic nuclei and scant cytoplasm. Glomeruloid structures are also present

Fig. 4 Blastema with tubules and surrounding mesenchyme





or teratomatous tissues. The lack of any teratomatous tissues and the absence of a renal mass ruled out the possibilities of a sacral teratoma and metastatic Wilms tumour, respectively. The final diagnosis, therefore, was a primary extrarenal Wilms tumour of the lumbosacral region.

Discussion

Heterotopic nonneoplastic renal tissue has been described in the heart, adrenal gland and lumbosacral region [2, 3, 11, 12, 14]. Heterotopic renal tissue in the heart and adrenal gland were incidental autopsy findings in adults [11, 12]. In contrast, heterotopic renal tissue in the lumbosacral region was seen in children [2, 3, 14]. One child also had a meningocele in the lower lumbar region [14]. Hence, heterotopic renal tissue in the lumbosacral region may occur with or without associated occult spinal dysraphisms. Cozzuto et al. [3], in explaining the presence of renal dysplasia in the sacral region, postulated that there is detachment of an embryonal focus originating from the metanephros with subsequent dysplastic growth. A similar reason was reiterated by Posalaky et al., who stated that these lesions might be due to misplaced remnants of meso- or metanephric tissue [14]. Differentiation of the metanephros into glomeruli and renal tubules is dependent on the stimulation of the branching ureteric bud. In heterotopic sites it has been suggested that there may be an abnormal source of stimulation or, alternatively, the metanephros may differentiate independently of an external stimulus (self-differentiation) [14]. Another theory is that these heterotopic renal foci may arise from normal or abnormal neural crest cells which have the potential for multicellular differentiation [14].

It is widely accepted that the classic intrarenal Wilms tumour, a common solid tumour of childhood, arises from the metanephric blastema [1]. In contrast, extrarenal Wilms tumours are rare and may occur at various sites: the retroperitoneum, cervix, ovary, uterus, inguinal/intrascrotal area and chest wall [16]. Extrarenal Wilms tumours pose challenging questions regarding their histogenesis. As a result, several histogenetic theories, often specific to the site of the tumour, have been postulated. Extrarenal Wilms tumours are generally believed to arise from heterotopic primitive renal tissue of either mesonephric or metanephric origin [16]. The mesonephros has the capacity for tubular and glomerular differentiation, but these structures usually disappear by the end of the 2nd month of embryological development [9] and the definitive kidney develops from the metanephros.

A review of cases recorded in the English language literature of Wilms tumours and immature renal tissue arising in association with spinal dysraphisms is shown in Table 1.

In conclusion, we have described a dorsal sacral tumour with extensive lumbosacral extradural extension, which is histologically consistent with an extrarenal Wilms tumour. Of special interest is the association with an undetected spinal dysraphism and the delay of 4 years before the onset of symptoms. Earlier, Posalaky et al. posed an important question of whether immature, heterotopic renal tissue in the lumbosacral area could be a precursor of Wilms tumour and concluded that delayed malignant transformation cannot be ruled out [14]. This case raises the same question, which is difficult to answer since it is impossible to know at what point malignant transformation occurred. It is not clear whether malignancy was present at birth or developed from heterotopic renal rests.

Note added in proof

Since the submission of this paper there has been a report of four cases of extrarenal nephrogenic proliferation in the sacrococcygeal region associated with spinal dysraphism. Two cases showed features of Wilms or incipient Wilms tumor.

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