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CASE REPORT
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COEXISTENCE OF SPINAL DYSRAPHISM AND EXTRARENAL WILMS TUMOUR: A CASE REPORT AND REVIEW OF THE LITERATURE

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ABSTRACT

Extrarenal Wilms tumor is rare, but its association with spinal dysfraism is even rarer, and to our knowledge, there is no association with meningomyelocele sac in the literature. Our case was a 36-month-old baby with a meningomyelocele sac in the lumbar region. No additional anomaly was detected, and the baby had no leg movements. Magnetic resonance imaging examination revealed a 24x10 mm meningomyelocele sac and an 18x30x27 mm contrast-forming mass in the sac. Mass resection was performed during sac repair. Histological examination revealed a three-phase tumor consisting of a stromal component containing fibrous and fatty tissue, an epithelial component containing primitive glomeruli and tubule structures consisting of papillary structures with fibrovascular nuclei within cystic structures, and a blastemal component containing oval primitive cells between epithelial areas. Immunohistochemically, the blastemal and epithelioid components were stained positively with WT1, the stromal component with vimentin, and the epithelial component with PanCK and EMA. In this form, the case was defined as an extrarenal Wilms tumor associated with a meningocolocele. This case was valuable in terms of showing that Wilms tumor should be included in the differential diagnosis of masses with meningomyelocele and the importance of total resection of these lesions.

Keywords: Extrarenal wins tumor, spinal dysraphism, meningomyelocele

INTRODUCTION

Wilms tumour is the most common primary kidney tumour in children, accounting for 5% of all childhood cancers[1,2]. Although the prognosis of Wilms tumour, which has three histological subtypes, varies according to stage, 4-year survival rates reportedly range from 85-100% for favourable histology, 70-100% for focal anaplastic type, and 30-35% for diffuse anaplastic type[3].

Extrarenal Wilms tumour is exceedingly rare. So much so that Shojaeian et al.[4], in their 2015 study in which they excluded cases coexisting with teratoma, stated that a total of 80 cases were published under the age of 14, the first of which was in 1961.

In patients with extrarenal Wilms tumour, localization is often the retroperitoneal, inguinal, lumbosacral regions, the genital organs, and mediastinum[4,5]. However, to our knowledge, the association of extrarenal Wilms tumour with spinal midline defect is very rare[6] and our literature review did not reveal any reported association with a meningomyelocele sac. In this case report, we present a case who had a postpartum mass with a meningomyelocele sac, which was later revealed to be a Wilms tumour as confirmed by pathological analysis.

CASE REPORT

A 36-week-old term female baby was evaluated because of a possible meningomyelocele sac in the lumbar region (Figure 1). There was no additional anomaly detected, and the infant did not have leg movements. Whole spinal and cranial magnetic resonance imaging (MRIs) were ordered to detect accompanying spinal pathologies in the patient who did not have cerebrospinal fluid (CSF) leakage from the meningomyelocele sac.

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Radiological Findings

In the evaluation of cranial and spinal MRIs, a defect in the posterior elements of the vertebrae in the lower thoracic-lumbar region, as well as a CSF-filled myelomeningocele sac measuring 24×10 mm characterized by neural elements in its lumen, were observed. At this level, abnormal osseous structures and fusion appearance in the vertebral bodies were recorded as additional findings. Finally, a mass lesion measuring 18×30×27 mm with lobulated contour and causing compression in the pouch was observed in the right lateral neighbourhood of the sac. It demonstrated heterogeneous features and relative hyperintensity in T2AGs, hypointensity in T1AGs, and was well-enhanced after intravenous gadolinium injection (Figure 2). In the differential diagnosis, meningioma was considered primarily with respect to imaging features and localization.

Urinary ultrasonography examination, which was performed later with regard to the primary tumour, did not reveal any remarkable findings, and the kidneys included in the MRI examination area were also natural in appearance.

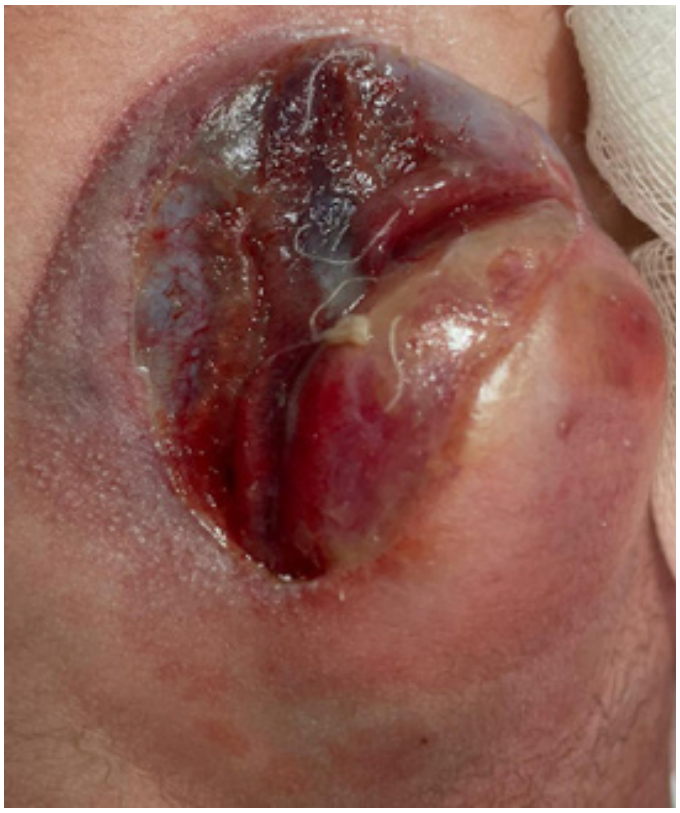

Figure 1. Preoperative view of the spinal defect

Surgical Intervention

The procedure was performed with the patient in the prone position. After total excision of the well-cleaved mass (Figure 3), the borders of which were clearly defined, the bone spur was excised and determined to be within the dura mater defect. Then, the duraplasty was completed by sectioning the filum, the skin was repaired, and the surgical intervention was completed.

Pathological Findings

Macroscopically, the specimen was observed to be a well-circumscribed lesion with an off-white fibrillary appearance, measuring 25×23×17 mm. Histological examination revealed a triphasic tumour consisting of a stromal component containing fibrous and adipose tissue, an epithelial component containing primitive glomeruli and tubule structures consisting of papillary structures with fibrovascular cores within cystic structures, and a blastemal component containing oval primitive cells between epithelial areas. There were no signs of anaplasia (Figure 4).

Immunohistochemically, the blastemal and epithelioid components were stained positively with WT1, the stromal component with vimentin, and the epithelial component with PanCK and EMA. The Ki-67 proliferation index was observed to be increased in focal areas.

Follow-up

The patient was started on chemotherapy and there are no long-term follow-up results as of writing.

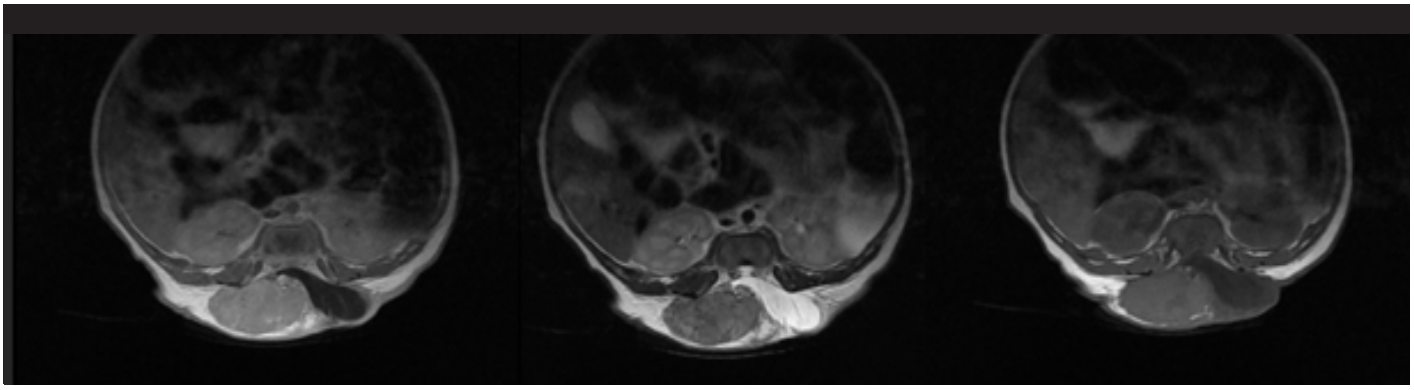

Figure 2. MRI image of the case

MRI: Magnetic resonance imaging

DISCUSSION

Extrarenal Wilms tumours are exceedingly rare, but the most common site is the retroperitoneum. Although it was previously reported with spinal midline defects such as diastematomyelia, tethered cord syndrome, and bone closure defects, to the best of our knowledge, this is the first report in the literature demonstrating an association with meningomyelocele.

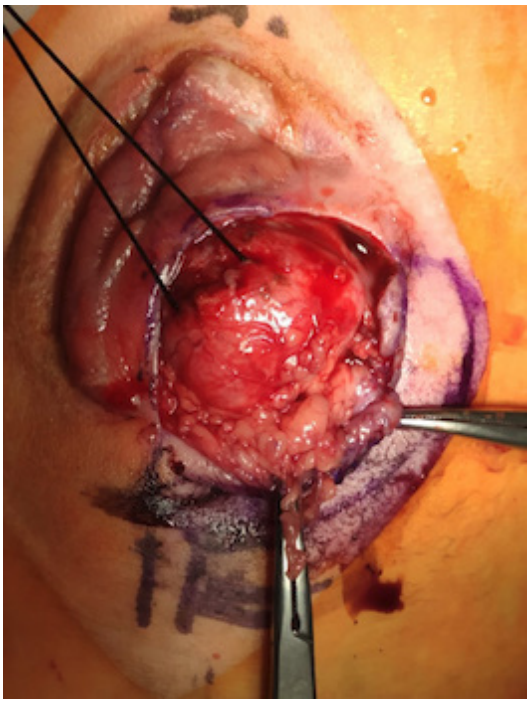

Figure 3. Perioperative view of the mass

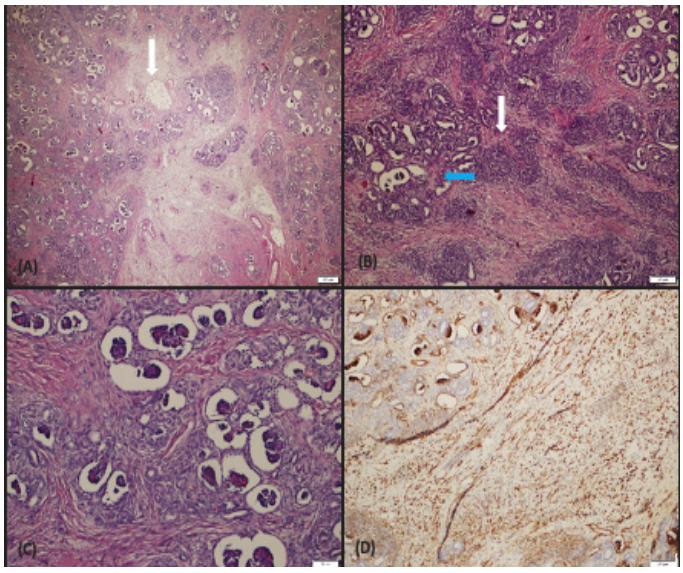

Figure 4. A: Mesenchymal component (arrow) containing loose myxoid stroma and adipose tissue, (H&E, x40), **B:** Blastemal (white arrow) and epithelioid (blue arrow) component (H&E, x100), **C:** Primitive glomerular structures formed by papillary structures containing fibrovascular cores within the cystic areas (H&E, x200), **D:** Positivity in epithelioid and blastemal component with WT-1 (x40)

Igbaseimokumo et al.[7], in their 2017 study in which they included patients with extrarenal Wilms tumour associated with spinal dysraphism, described a total of seven cases (together with their own). When these cases were examined, it was found that five cases had swelling in the lumbar region accompanied by hypertrichosis, while there were no morphological changes in the other two cases. Among these seven patients reported by the authors, we did not include the cases reported by Deshpande et al.[13] and Fahner et al.[14] in the present study because they did not have any anomalies that could be considered as spinal dysraphism. In addition, in the review of Govender et al.[9] one of the cases was mentioned as a teratocarcinoma and the outcome pathology was evaluated as Wilms tumour; however, we excluded this case because it was considered controversial in its reportedstate. Besides, unlike these, in the study of four cases titled, "Extrarenal Nephroblastic Proliferation in Spinal Dysraphism" presented by Abrahams et al.[10], one of the cases was a 4-week-old girl with a bifid lamina in the L5 vertebra and a 35×35 mm mass in this region, and therefore, was included in our assessment because the mass pathology was Wilms tumour. To conclude, we defined our case as the seventh case adding to a total of previously-reported six patients with extrarenal Wilms tumour associated with spinal dysraphism, and we believe the association with a meningomyelocele sac in our study was a valuable finding (Table 1).

The generally accepted view regarding Wilms tumour occurrence outside of renal tissue is that mesenchymal remnants transform into nephrogenic remnants, which in turn undergo malignant transformation[15-17].

Wilms tumour has no specific radiological features, and therefore, pathological examination is critical. When other cases in the literature are examined, it is understood that in the preoperative evaluation of extrarenal Wilms tumours, other diagnoses are considered possible by physicians with respect to the patient's age, clinical findings, and tumour localization[7]. In addition, neuroblastoma, teratoma, embryonal rhabdomyosarcoma and other rare sarcomas are included in the radiological differential diagnosis of extrarenal Wilms tumours[5].

However, in the pathological examination of our case, the tumour was considered compatible with the diagnosis of Wilms tumour without anaplasia, since the tumour had triphasic morphology including blastemal, epithelial, and mesenchymal components, was immunohistochemically positive for WT-1, had a blastemal component, was immunohistochemically positive for WT-1, and did not have a typical teratoma area.

Since there is no specific radiological feature of Wilms tumour and the diagnosis can only be made pathologically, it should be considered in the differential diagnosis when a mass lesion accompanying meningomyelocele is encountered.

Table 1. Literature of extrarenal Wilms tumour cases associated with spinal dysraphism and their characteristics

					Treatment			
Study	**Age**	**Sex**	**Level**	**Spinal dysraphism**	**Surgery**	**ChT**	**RT**	**Follow-up**
Present case	New-born	Female	T12-L5	Meningomyelocele	Complete excision	Yes	No	2 months
Igbaseimoku et al.[7] 2017	New-born	Female	L5	Dysraphic lamina + lipoma	Complete excision	Yes	No	No recurrence at 1 year
Sharma et al.[8] 2005	18 months	Female	L1	Diastematomyelia + lipoma	Complete excision	Yes	No	3 months
Govender et al.[9] 2000	4 years	Female	T10-coccyx	Spina Bifida	Biopsy	Yes	Yes	Palliative care
Abrahams et al.[10] 1999	4 weeks	Female	L5	Bifid lamina	Complete excision	Yes	No	4 years disease-free after 10 weeks of chemotherapy
Mirkin et al.[11] 1990	2 years	Female	T12-L4	Diastematomyelia + lipoma	Complete excision	Yes	Yes	Cerebellar metastasis 1 year after diagnosis, no recurrence 20 months after cerebellar mass excision
Fernbach et al.[12] 1984	2 years	Female	L1	Diastematomyelia + lipoma	Near total excision	Yes	Yes	No recurrence at 1 year

ChT: Chemotherapy, RT: Radiotherapy

Ethics

Informed Consent: Written informed consent was obtained.

Authorship Contributions

Surgical and Medical Practices: A.T., T.T., B.Y., Z.Ç.G., Y.Ç., Ö.Ö., Concept: A.T., T.T., Design: A.T., T.T., Data Collection or Processing: B.Y., Z.Ç.G., Analysis or Interpretation: A.T., Y.Ç., Ö.Ö., Literature Search: A.T., T.T., B.Y., Z.Ç.G., Y.Ç., Ö.Ö., Writing: A.T., T.T., Y.Ç., Ö.Ö.

Conflict of Interest: The authors have no conflicts of interest to declare.

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