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Unusual Presentation of Wilms' Tumor in a 4-Month-Old Infant as Presternal Metastatic Swelling—A Case Report with Review of Literature

Md Fahim Ahmad[1] Prema Menon[1] Shailesh Solanki[1] Amita Trehan[2] Parikshaa Gupta[3]

[1] Department of Pediatric Surgery, Postgraduate Institute of Medical Education and Research, Chandigarh, India
[2] Department of Pediatric Hemato-oncology, Postgraduate Institute of Medical Education and Research, Chandigarh, India
[3] Department of Cytology and Gynaecologic Pathology, Postgraduate Institute of Medical Education and Research, Chandigarh, India

Address for correspondence Prema Menon, MS, DNB, FRCS, MCh, FMAS, Department of Pediatric Surgery, Advanced Pediatrics Centre, Postgraduate Institute of Medical Education and Research, Room No. 3133, Level 3-A, Sector 12, Chandigarh 160012, India (e-mail: menonprema@hotmail.com).

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Abstract

A 4-month-old infant presented with an enlarging large vascular presternal swelling noticed for the past 2 months. Clinical examination revealed a left renal mass in this otherwise asymptomatic child. She underwent left nephroureterectomy and excision of the skin metastasis following a course of chemotherapy. Preoperative cytology and postoperative histopathological examination confirmed Wilms' tumor with a single skin metastasis. We report this case for its rarity.

Keywords

- Wilms' tumor
- cutaneous metastasis
- presternal swelling
- anaplasia

Introduction

Wilms' tumor is the most common primary renal malignancy of childhood. It constitutes 6.3% of cases of childhood cancer and accounts for approximately 90% of all pediatric renal tumors.[1] Tumor usually arises from single kidney; however, there can be synchronous or multifocal tumors in around 10% of the patients and these usually tends to present at an earlier age. It is usually seen in children aged between 3 and 5 years and is unusual before 6 months of age.[2] The vast majority of patients presents with asymptomatic abdominal mass. In one-third of patients, there can be abdominal pain, hematuria, and hypertension. Rarely there can be atypical presentation because of compression of surrounding organs or infiltration into renal vein and inferior vena cava.[3] Lung is the most common site of metastasis followed by liver and contralateral kidney.[4] Cutaneous manifestations are not common in Wilms' tumor unlike other tumors like neuroblastoma, leukemia, rhabdoid tumor, and rhabdomyosarcoma that can present in early infancy with metastasis.[5]

In the index case, a 4-month-old infant presented with a presternal swelling and on further examination was found to have a left-sided flank mass. The diagnosis of Wilms' tumor with cutaneous metastasis was confirmed after further workup.

Case Report

A 4-month-old female child, first in birth order, born by full-term vaginal delivery, and asymptomatic at birth, was referred with a presternal swelling. This had been noticed since the age of 2 months by the parents and was gradually increasing in size. There was no history of loss of appetite or weight. She had normal bowel and bladder habits. There were no complaints other than the presence of the presternal swelling. An ultrasonography (USG) of the swelling done before referral was suggestive of hemangioma or arteriovenous malformation. On examination, a 6×4 cm, round, well defined, firm, nontender, noncompressible swelling was present over the presternal area with overlying darkened skin and few areas of ulceration and eschar (►**Fig. 1A**).

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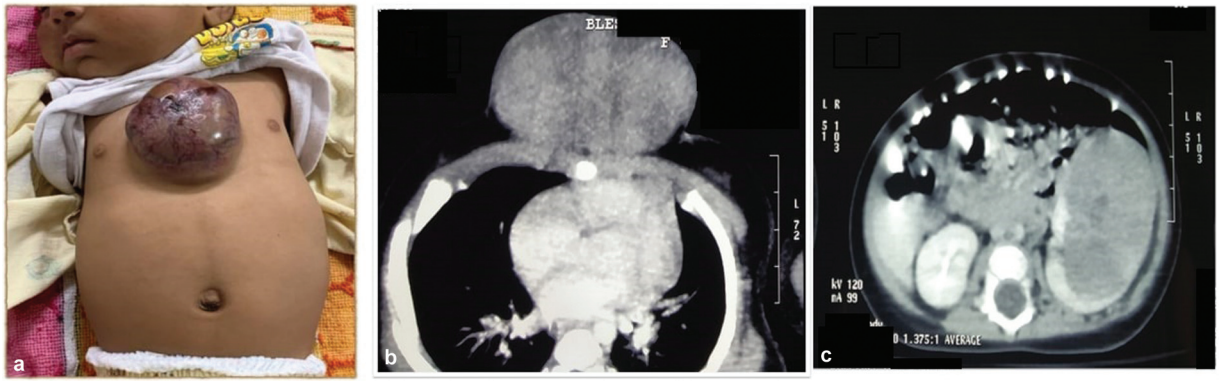

Fig. 1 (**A**) Single large skin metastasis in lower presternal area at presentation; (**B**) contrast-enhanced computed tomographic scan of chest shows a $3.8 \times 3.8 \times 6.3$ cm mass in the presternal area lying in subcutaneous plane with no intrathoracic extension; (**C**) contrast-enhanced computed tomographic scan of abdomen shows a $5.1 \times 3.8 \times 5.3$ cm hyperdense exophytic mass arising from the left kidney with no major vessel encasement or calcifications.

Incidentally, fullness was noted in the left flank region and on abdominal examination, a mass of size 4×5 cm was palpable in the left lumbar region that was firm, well defined, smooth, bimanually palpable, and nonballotable. She had no features of any syndrome associated with Wilms' tumor. USG abdomen revealed a hyperechoic 4×4 cm left renal mass. Contrast-enhanced computed tomography (CECT) chest showed that the presternal mass had no intrathoracic extension. (►**Fig. 1B**). CECT of abdomen showed a $5.1 \times 3.8 \times 5.3$ cm hyperdense exophytic mass arising from the left kidney with no major vessel encasement or calcifications (►**Fig. 1C**).

Fine-needle aspiration cytology smears from the left renal mass and chest wall swelling showed a predominantly dispersed population of blastemal cells with high nucleocytoplasmic ratio and hyperchromatic nuclei. Focal anaplasia and occasional mesenchymal fragments could also be noted. Cell blocks with immunohistochemistry were also suggestive of anaplastic Wilms' tumor with metastasis (►**Fig. 2**). Both the presternal swelling and renal mass showed a reduction in size after four cycles of age adjusted chemotherapy with doxorubicin, actinomycin D, and vincristine. The patient underwent left radical nephroureterectomy with lymph node sampling and excision of the chest wall lesion. Histopathological examination confirmed Wilms' tumor with lymph nodes free of tumor. Local staging was stage 1 as the tumor was fully excised and there was 99% necrosis. Margins of the local site tumor (abdomen) and metastatic site were also negative. After surgery, she developed surgical site infection that responded to antibiotics. The tumor was triphasic in nature and there was focal anaplasia on the initial biopsy that does not qualify for unfavorable histology. She therefore did not belong to poor risk histology and was planned for 27 weeks of chemotherapy with actinomycin D, doxorubicin, and vincristine. She defaulted after receiving 12 cycles of chemotherapy. The reason for abandonment of therapy is unclear. She expired 6 months after surgery at home after a brief period of complaint of fever, weight loss, and cough. The probable cause of death may be due to coronavirus disease 2019 or the malignancy itself.

Discussion

Wilms' tumor is the second most common childhood abdominal malignancy forming 6% of all the childhood cancers with a 5-year survival rate of nearly 90%.[1]

It usually presents as an asymptomatic abdominal mass. Other presentations include abdominal pain, fever, hematuria, hypertension, recurrent urinary tract infection, and anemia.[2,3] Metastasis occurs commonly in the lung, liver, and contralateral kidney. Less common sites include bone, skin, brain and orbit, and rarely testes.[4] Wilms' tumor is less frequent in infants with the mean age at diagnosis being around 8 months.[2] Complete surgical removal without tumor spillage is essential as these patients have sixfold increased risk of local abdominal relapse.[6] Because of refinement in risk stratification and advancement in chemotherapy, the overall survival has improved to greater than 90% for localized disease and 75% for metastatic disease.[7]

Unlike in older children, Wilms' tumor in infancy often has cystic appearance.[8] This cystic form is considered to be low-risk nephroblastoma, prognosis is better, and they may be treated with surgical resection alone.[9] Early detection is possible in specific syndromic associations if frequent checkups are done in the first year of life. In our case, the child presented at 4 months of age with cutaneous metastasis and features of anaplasia on cytological examination. Other more malignant tumors could be ruled out by immunohistochemistry of cell blocks, namely: malignant rhabdoid tumor kidney (as INI-1 retained), clear cell sarcoma kidney (cyclinD1 negative), neuroblastoma (chromogranin and synaptophysin negative), Ewing's sarcoma (CD99 negative), and lymphoma (leukocyte common antigen negative). The tumor cells were positive for vimentin and desmin.

Currently, tumor histology is considered as the most common "biomarker" reflecting prognosis. Anaplastic histology, especially diffuse anaplasia, is associated with higher recurrence rates, metastases, and death. The recommended National Wilms' Tumor Study group/Children's Oncology group chemotherapy protocols differentiate patients by presence and degree of anaplasia.[10,11]

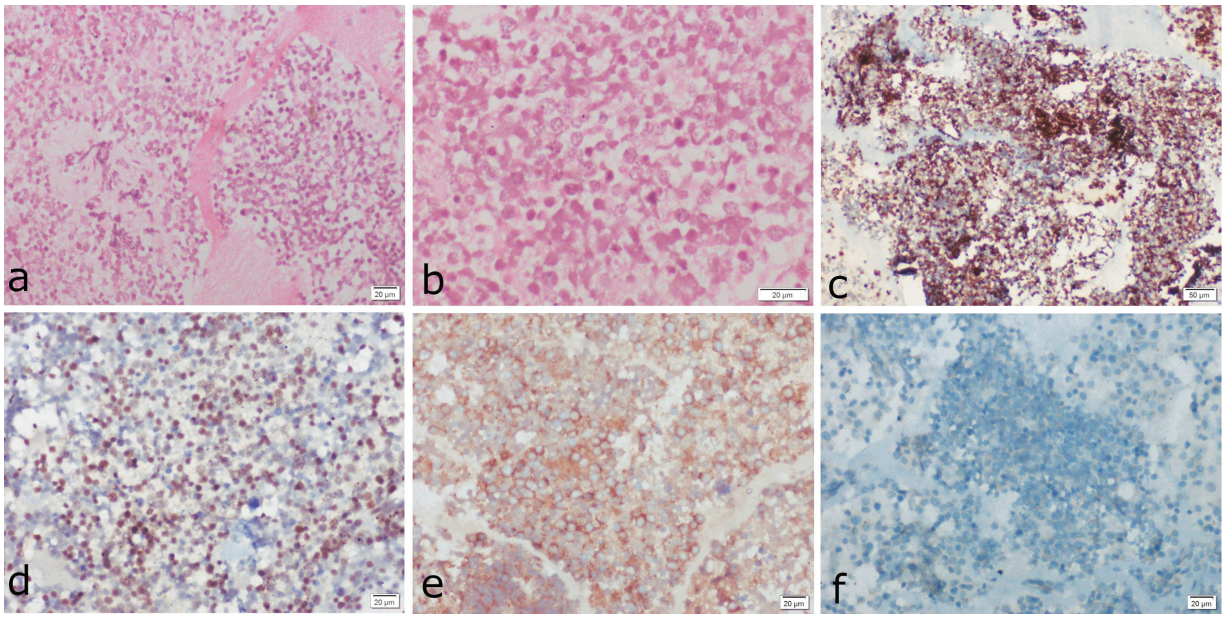

Fig. 2 (**A**, **B**) Sections from the cell block from the left renal mass and chest wall swelling showing a dispersed population and sheets of blastemal cells and occasional mesenchymal fragments (hematoxylin and eosin; **A**:20x, **B**:40x); (**C**–**F**) Immunocytochemistry for vimentin (**C**: 10x) shows diffuse strong positivity in the tumor cells, retained nuclear expression for INI-1 (**D**: 20x), and dot-like positivity for desmin (**E**: 20x). The tumor cells were negative for chromogranin, synaptophysin, WT1 (**F**: 20x), CD45, cyclinD1, and CD99.

On reviewing literature, we did not find any case of Wilms' tumor presenting as a single skin metastasis in an infant. We found one report of a 12-year-old child who had undergone nephrectomy 7 years back for Wilms' tumor, and presented with a lump in the parasternal area as a form of late recurrence.[12]

Conclusion

This case reveals an unusual presentation of Wilms' tumor in an infant that has not been reported before in literature. The child manifested first with a skin lesion at 2 months of age and on histopathological examination was revealed to have anaplastic Wilms' tumor, both of which are extremely rare.

Conflict of Interest
None declared.

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