Case Report

# Malignant extrarenal rhabdoid tumor of parapharyngeal space: A rare presentation

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**Abstract** Malignant extrarenal rhabdoid tumors (ERRTs) are very rare, highly aggressive tumors and are associated with a poor prognosis. They usually occur in young children. Diligent histopathological examination along with the use of judicious immunohistochemistry (IHC) panel is essential to diagnose this rare entity with distinct therapeutic implications. We report a rare case of ERRT with extension up to the skull base involving the right jugular fossa with thrombus in the right internal jugular vein.

**Keywords:** Extrarenal rhabdoid tumor, INI expression, parapharygeal space

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## INTRODUCTION

Malignant extrarenal rhabdoid tumor (ERRT) is an extremely infrequent, aggressive, and high-grade tumor. The tumor is usually observed in infants and very young children. Important differential diagnoses include rhabdomyosarcoma (RMS), lymphoma, sarcoma with epithelioid differentiation, and melanoma. The histogenesis is uncertain; on microscopy, characteristic features of this tumor are diffuse proliferation, round or polygonal "rhabdoid cells" with eccentric nuclei, prominent nucleoli, and glassy eosinophilic cytoplasm with hyaline-like inclusion bodies. On immunohistochemistry, it exhibits epithelial as well as mesenchymal differentiation.

## CASE REPORT

A 4-year-old boy with no significant past and family history presented with the complaints of pain and swelling over the right side of the neck radiating to the right ear for 2 months associated with low-grade fever, loss of weight, and appetite. No history of bone pain, joint pain, or jaundice was found.

On local examination, there was a swelling in the right upper neck of size 3 cm × 4 cm. It was firm, nontender, immobile, and extending into the submandibular region with palpable subcentimeter left cervical lymph nodes. Systemic examination was normal.

Routine laboratory hematological and biochemical investigations were unremarkable. Ultrasonography neck revealed 5.5 cm × 4 cm solid mass with internal vascularity at right upper lateral neck. Contrast-enhanced computed tomography (CECT) neck revealed heterogeneously enhancing lesion (5.6 cm × 3.3 cm) in the right

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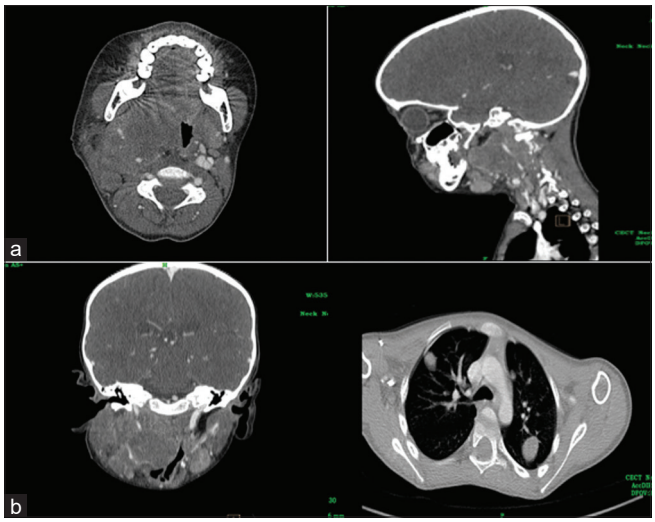

**Figure 1:** (a) Contrast-enhanced computed tomography neck reveals heterogeneous enhancing lesion (5.6 cm × 3.3 cm) in the right parapharyngeal space compressing/encasing the adjacent structure. (b) Contrast-enhanced computed tomography chest shows multiple nodular lesions in both lungs, suggestive of tumor infiltrates

parapharyngeal space compressing and encasing the adjacent structures [Figure 1a]. CECT chest showed multiple nodular lesions in both lungs, suggestive of tumor infiltrates [Figure 1b]. CECT brain was done in view of persistent headache, which showed that parapharyngeal mass is extending up to the skull base involving the right jugular fossa with thrombus in the right internal jugular vein.

On the basis of clinical and radiological findings, a provisional diagnosis of lymphoma was considered. Fine-needle aspiration cytology smears were suggestive of undifferentiated malignancy favoring non-Hodgkin's lymphoma. Right parapharyngeal mass biopsy was done which revealed a diffuse sheet of large neoplastic cells which are plump spindle to round having vesicular nucleus and prominent nucleoli with moderate amount of dense eosinophilic cytoplasm [Figure 2a]. Brisk mitosis was evident, and necrosis was also seen [Figure 2b].

Histomorphologically, it was an undifferentiated malignancy with differentials in the following order: RMS, lymphoma, epithelioid sarcoma, and melanoma. Primary IHC panel was performed including cytokeratin (CK), leukocyte common antigen, desmin, myogenin, terminal deoxynucleotide transferase, epithelial membrane antigen (EMA), and S-100, of which only CK with inclusion like positivity and vimentin was expressed which pointed toward sarcoma with epithelioid differentiation and a rare possibility of undifferentiated carcinoma, while ruling out other differentials. Further, IHC panel was given which showed focal positivity for smooth muscle actin, whereas negativity

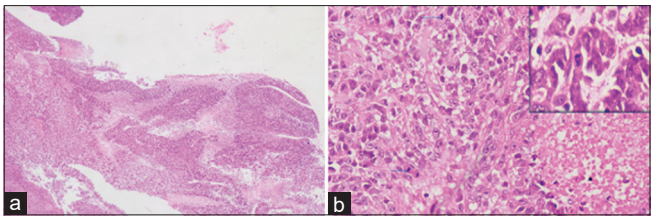

**Figure 2:** (a) Tumor cells in sheets with peritheliomatous pattern and foci of necrosis (H and E, ×40). (b) Diffuse sheets of neoplastic cells which were plump spindle to round having vesicular nucleus and prominent nucleoli with moderate amount of eosinophilic cytoplasm (H and E, ×400). Arrow shows mitotic figure

for CK5, P40, CK7, and synaptophysin. integrase interactor 1 (INI- 1) expression was lost [Figure 3] in tumor cells. MIB 1 labelling index/Ki67 (MIB-1) index was approximately 80% [Figure 3]. Clinical profile espoused with histopathological and IHC findings provided a tangible evidence to render a final diagnosis of ERRT.

On further workup, bone marrow aspiration and biopsy were free of tumor. The patient was started on ifosfamide-, cisplatin-, and etoposide-based chemotherapy, but he could not tolerate chemotherapy. He developed recurrent vomiting and tonic-clonic seizures, leading to dyselectrolemia and severe hyponatremia and succumbed to his illness in 2 weeks.

## DISCUSSION

Malignant rhabdoid tumor (MRT) has been originally described in the kidney and has been reported as "rhabdomyosarcomatous variant of Wilms tumor,"[1] but subsequently has been reported in various extrarenal sites also. Now, ERRT has been defined as a distinct clinicopathologic entity with a distinct clinical course and histopathologic and IHC findings and cytogenetics.[1-3]

The usual age of presentation is below 5 years. The incidence is 0.15/million under 15 years age.[4] No predominance regarding gender or ethnicity has been observed. Central nervous system is the most frequent site; however, it has been reported in other sites also, such as liver, brain, tongue, neck, chest, pelvis, extremities, and several other sites.[2] To our knowledge, this is the second reported case of ERRT in parapharyngeal space, with the first case being reported by Sparano *et al*.[3]

Microscopic analysis shows the presence of a monotonous population of large, round polygonal cells with abundant eosinophilic cytoplasm, vesicular eccentric nuclei, and prominent nucleoli.[5] The most striking morphological feature is the deeply, homogeneously eosinophilic cytoplasm of the tumor cells, with eccentric nucleus.

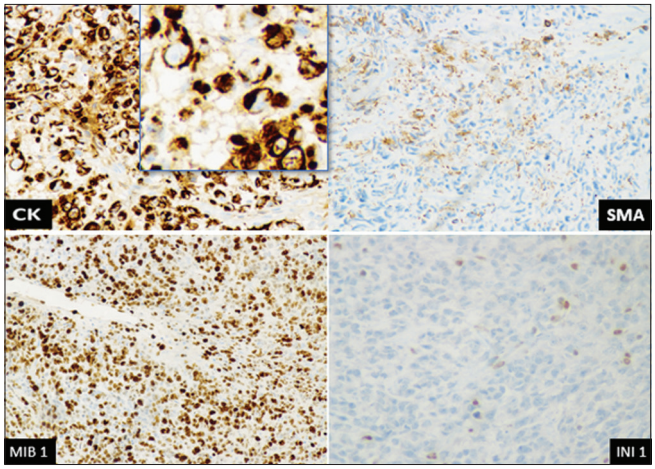


**Figure 3:** On IHC, tumor cells were positive for cytokeratin (inclusion like positivity) and focally positive for smooth muscle actin. MIB-1 index was approximately 80%. INI expression was absent

Differential diagnosis in children includes ERRT, RMS, and lymphoma, whereas in adults, it includes epithelioid sarcoma, RMS, epithelioid malignant peripheral nerve sheath tumor (MPNST), and melanoma.

Immunohistochemically, there is positivity for vimentin, often for keratin and EMA, and rarely for desmin, although generally not for skeletal muscle markers (Myogen/MyoD1) or S-100 protein.

Defining feature of ERRTs is an aberration (mutation or deletion) of the INI1/SMARCB1 gene located at chromosome 22q11.1.[6] However, rare MRTs are also SMARCA4 deficient. SMARCB1 gene (INI-1, BAF47) is a member of the SWItch/Sucrose nonfermentable chromatin remodeling complex, involved in the epigenetic regulation of gene transcription. SMARCB1 acts as a tumor suppressor gene, and loss of function of both alleles gives rise to SMARCB1-deficient tumors which include MRT, atypical teratoid/ teratoid/ rhabdoid tumor (AT/RT), cribriform neuroepithelial tumor, renal medullary carcinoma, and epithelioid sarcoma. Other tumors including subsets of epithelioid MPNST, subsets of chordomas, myoepithelial carcinomas, Schwannomas arising in Schwannomatosis, and sinonasal carcinomas have been associated with variable loss of SMARCB1 expression. Variable and reduced expression of SMARCB1 is characteristic of synovial sarcoma.[7]

ERRT is a highly aggressive tumor and may metastasize to the lungs, liver, and lymph nodes. Surgery followed by adjuvant chemotherapy and radiotherapy is the preferred management. However, most of the cases present in advanced stage and are subjected to only chemotherapy and/or radiotherapy. In spite of multimodality treatment, the prognosis is dismal.

## CONCLUSION

ERRTs are a heterogeneous group of aggressive tumors with distinct histopathological and IHC findings. Diligent histopathological examination and judicious use of IHC panel in correlation with clinical profile will guide in accurate diagnosis as it carries distinct therapeutic and prognostic implications.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

### Conflicts of interest

There are no conflicts of interest.

## REFERENCES

1. Beckwith JB, Palmer NF. Histopathology and prognosis of Wilms tumors: Results from the first national wilms' tumor study. Cancer 1978;41:1937-48.
2. Mazzocchi M, Chiummariello S, Bistoni G, Marchetti F, Alfano C. Extrarenal malignant rhabdoid tumour of the heel – A case report. Anticancer Res 2005;25:4573-6.
3. Sparano A, Kreiger P, Kazahaya K. Malignant rhabdoid tumor of the parapharyngeal space. Ear Nose Throat J 2009;88:E24-6.
4. Bourdeaut F, Fréneaux P, Thuille B, Bergeron C, Laurence V, Brugières L, *et al*. Extra-renal non-cerebral rhabdoid tumours. Pediatr Blood Cancer 2008;51:363-8.
5. Fanburg-Smith JC, Hengge M, Hengge UR, Smith JS Jr., Miettinen M. Extrarenal rhabdoid tumors of soft tissue: A clinicopathologic and immunohistochemical study of 18 cases. Ann Diagn Pathol 1998;2:351-62.
6. Hoot AC, Russo P, Judkins AR, Perlman EJ, Biegel JA. Immunohistochemical analysis of hSNF5/INI1 distinguishes renal and extra-renal malignant rhabdoid tumors from other pediatric soft tissue tumors. Am J Surg Pathol 2004;28:1485-91.
7. Pawel BR. SMARCB1-deficient tumors of childhood: A Practical guide. Pediatr Dev Pathol 2018;21:6-28.