# Case of the month
# November 2018

## Malignant extra-renal rhabdoid tumour of right parapharyngeal space: A rare presentation

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# INTRODUCTION

- Malignant extra-renal rhabdoid tumours :- very rare but highly aggressive tumour.

- Difficult to diagnose because of its varied morphological features

# Case Presentation

- 4 year old boy presented with
  - complaints of pain and swelling over right side of neck for 2 months
  - Pain referred to right side of ear
- Associated with fever, weight loss and anorexia.
- No h/o bone pains, jaundice, bleeding manifestation, blood transfusion or steroid intake.
- O/E -patient was irritable, afebrile, swelling in right upper neck was 3x4cm, firm, non tender, immobile and extending upto submandibular region.

# INVESTIGATIONS

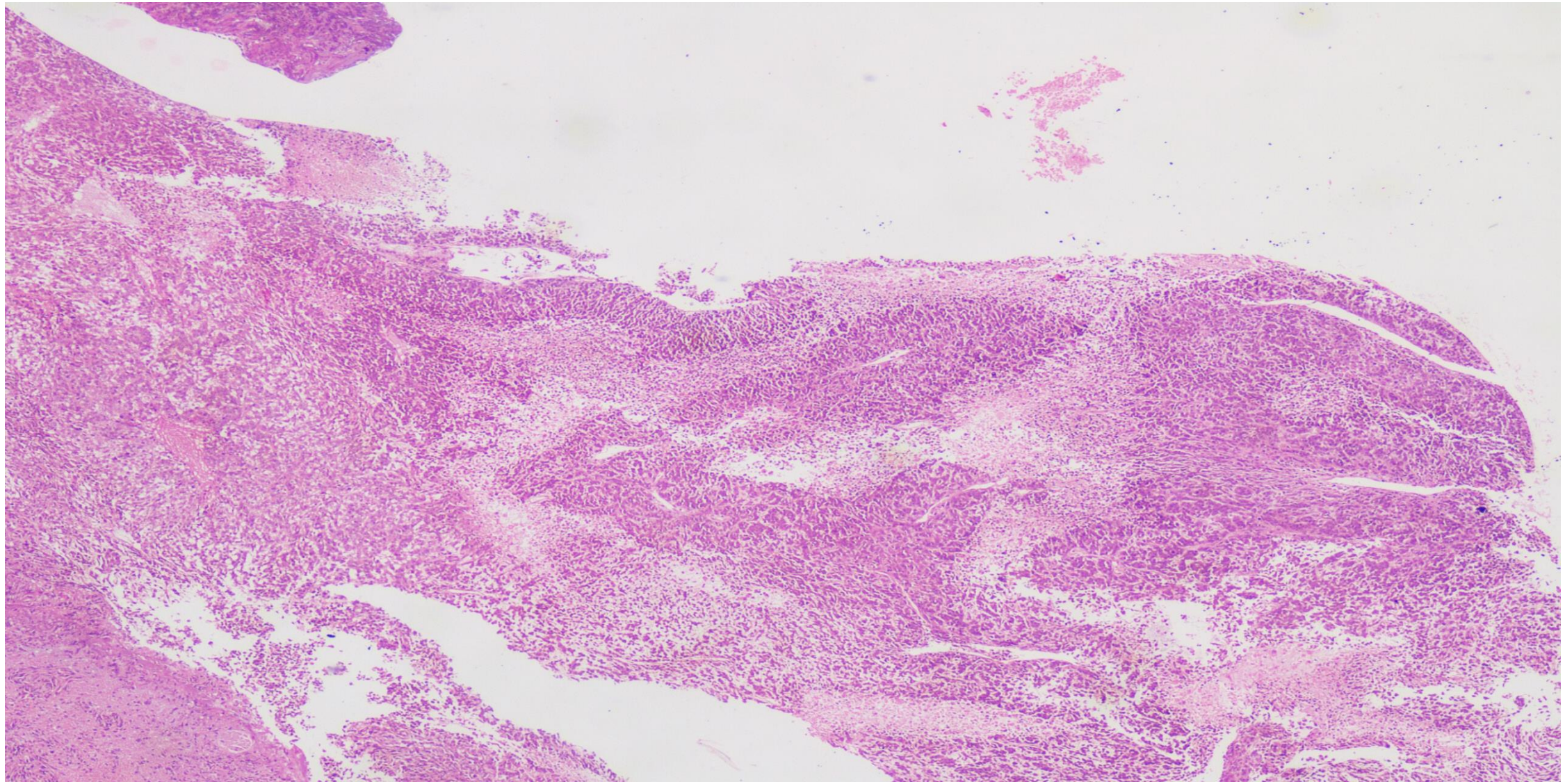
- **USG neck** revealed right upper lateral neck solid appearing lesion(5.5x4cm) with internal vascularity.

- **CECT neck and chest** revealed heterogeneous enhancing lesion (5.6 X 3.3 cm) in right parapharyngeal space compressing / encasing adjacent structure with multiple nodular lesions in both lungs

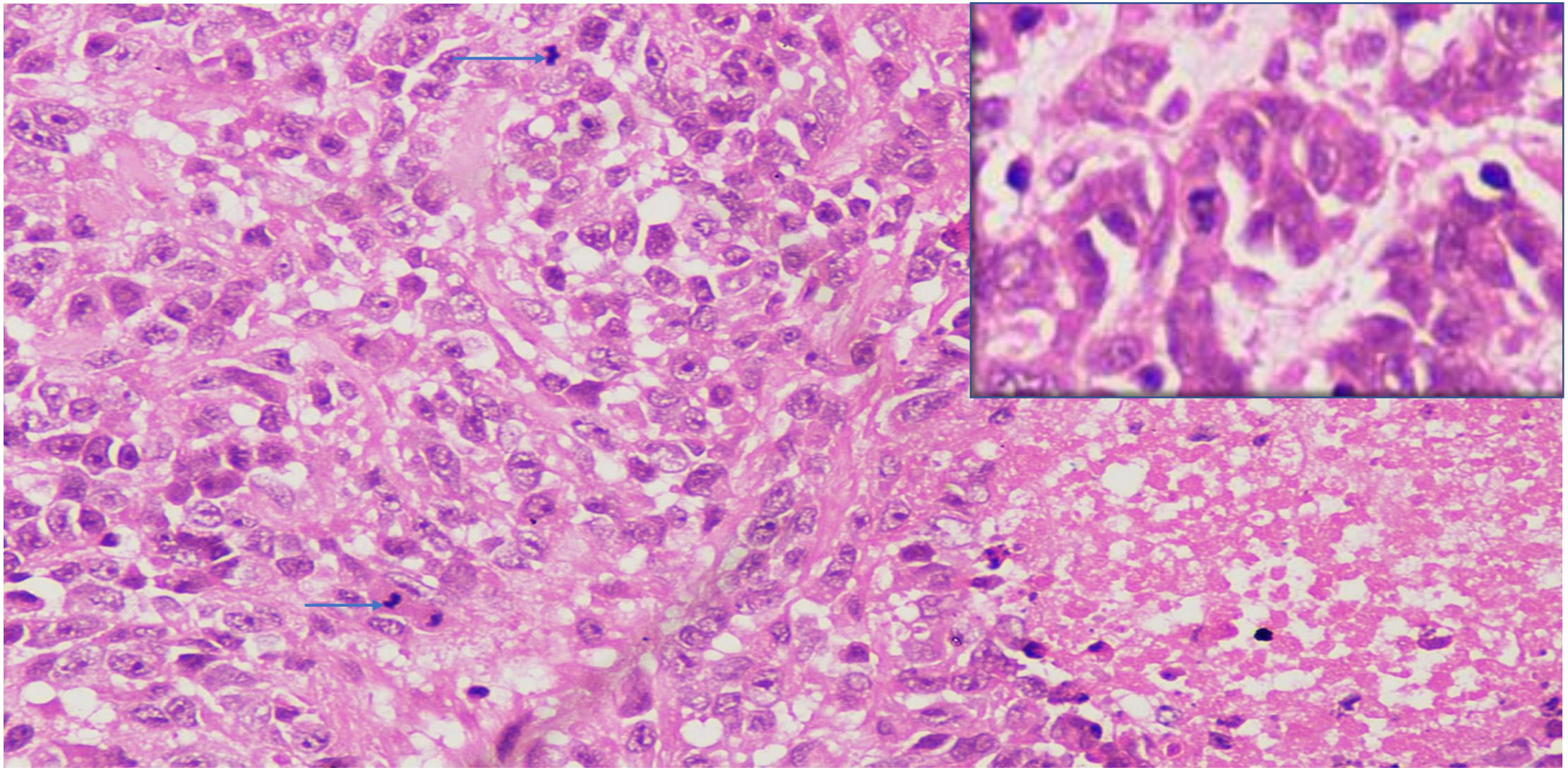
- Right cervical lymph node biopsy was done.

- On microscopic examination -
  - Sections revealed effaced lymph node architecture comprising of diffuse sheets of neoplastic cells which were plump spindle to round, having vesicular nucleus, prominent nucleoli with moderate amount of eosinophilic cytoplasm.
  - Brisk mitosis was evident and necrosis was also seen.

**H&E (40x):** Tumor cells in sheets with peritheliomatous pattern and foci of necrosis



**H&E (400X):** shows diffuse sheets of neoplastic cells which were plump spindle to round having vesicular nucleus, prominent nucleoli with moderate amount of eosinophilic cytoplasm. Arrow shows mitotic figures



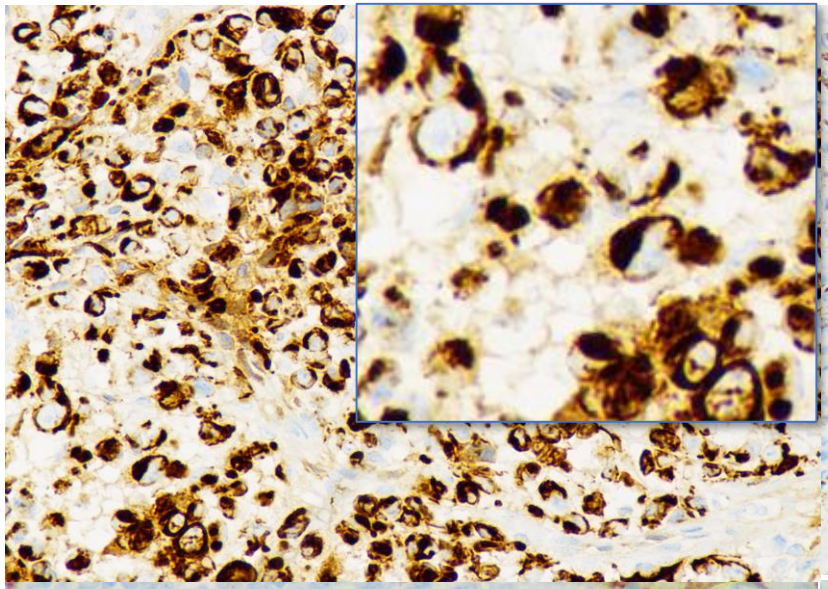
# Histomorphological DD

- Rhabdomyosarcoma
- Lymphoma
- Sarcoma with epithelioid differentiation
- Melanoma

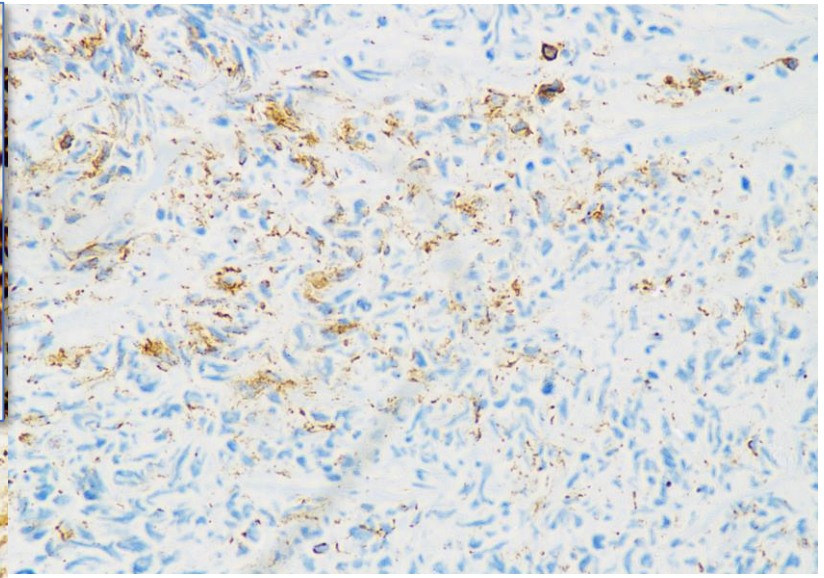
# IMMUNOHISTOCHEMISTRY

- Tumour cells revealed common mesenchymal and epithelial differentiation.
  - Tumor cells were positive for CK (inclusion like positivity),
  - Focally positive for SMA while
  - Negative for LCA, Desmin, myogenin, Tdt, EMA, S-100, Synaptophysin, CK5 and p40.

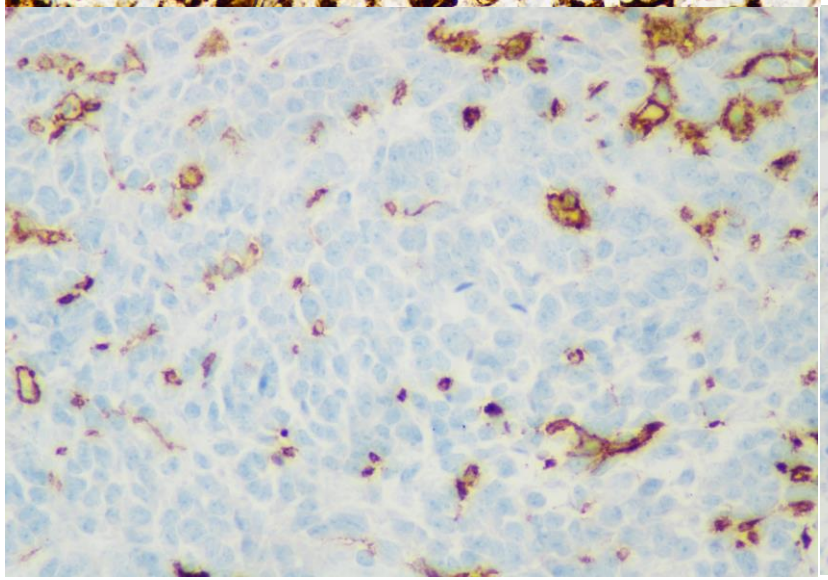
CK



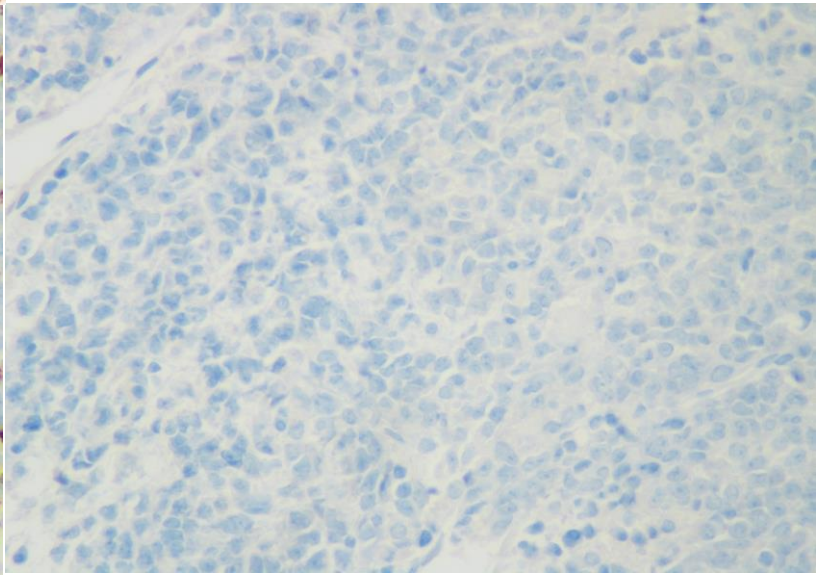
SMA



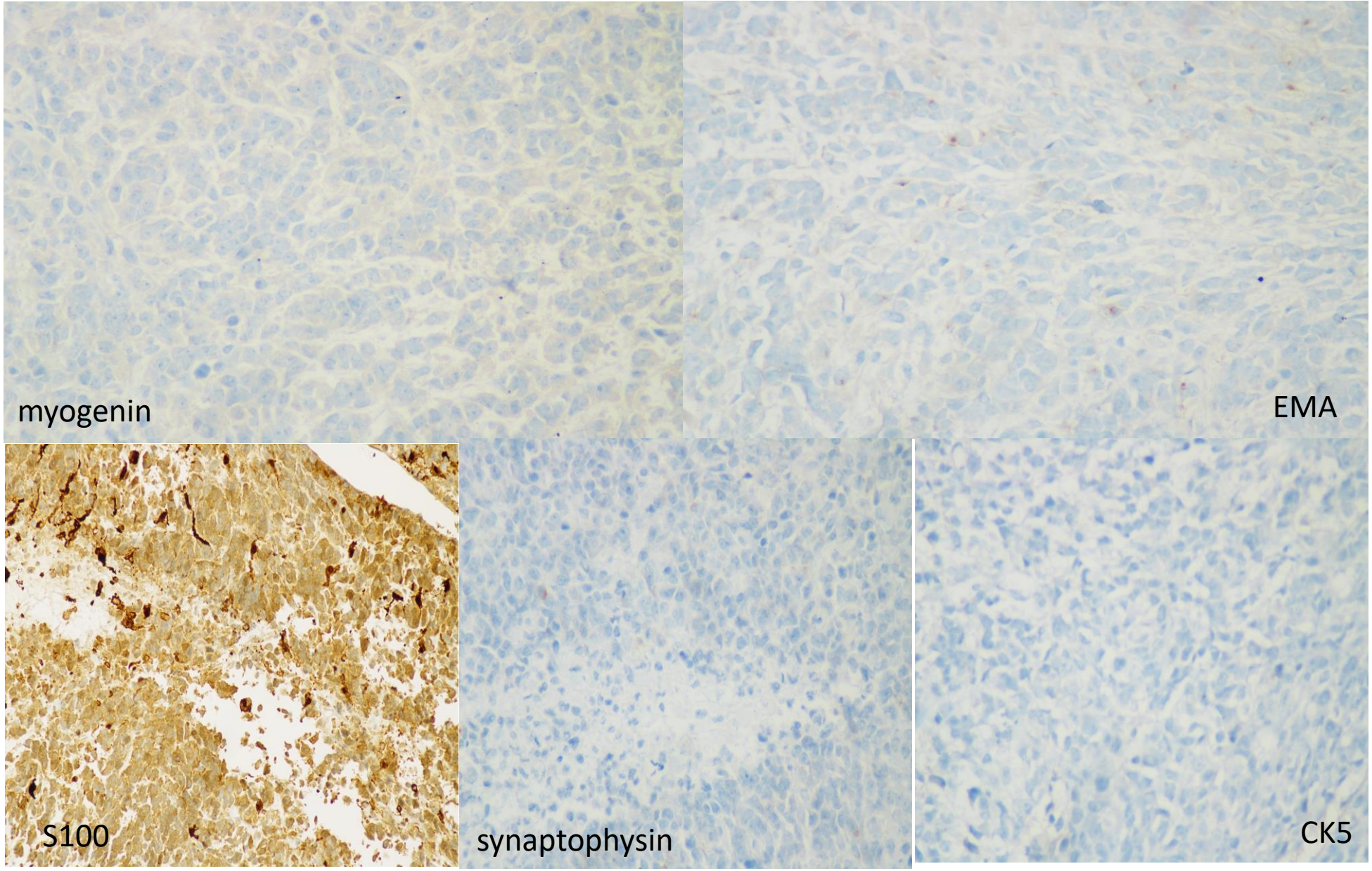
LCA



DESMIN

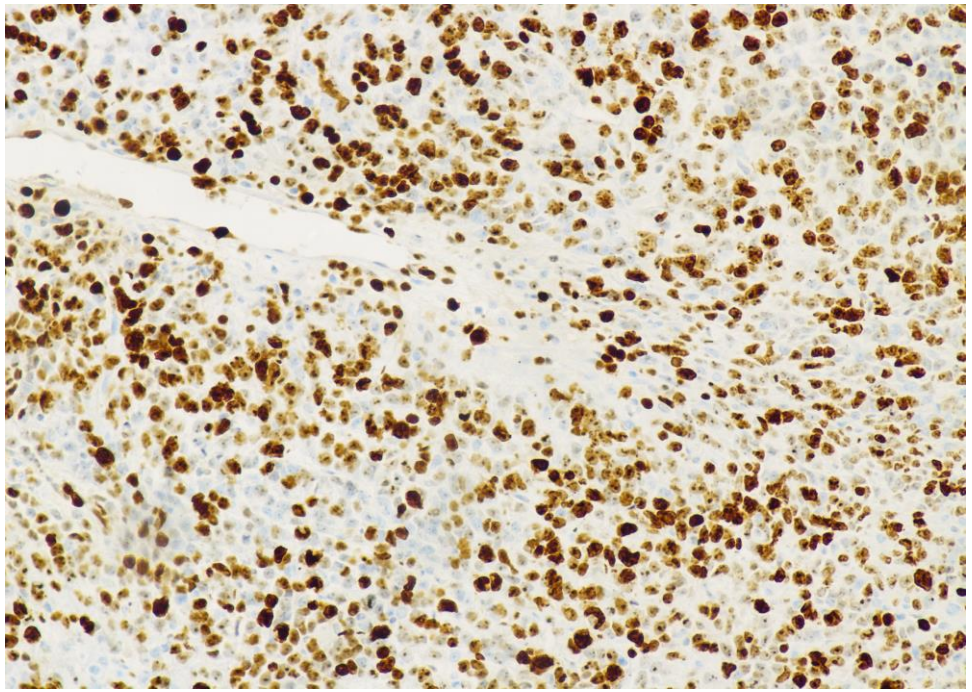


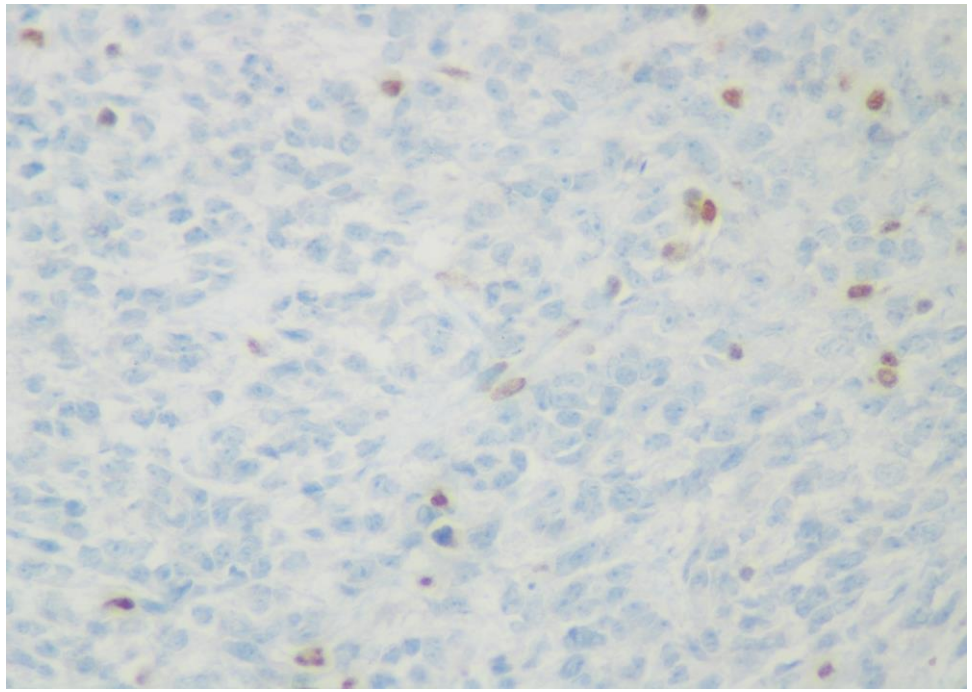
On IHC tumor cells were positive for CK (inclusion like positivity as shown in inset), and focally positive for SMA . negative for LCA, Desmin.




Tumor cells negative for myogenin, EMA, S-100, Synaptophysin and CK5

MIB 1





INI 1

MIB-1 index was approx. 80%.

INI expression was absent.

# FINAL DIAGNOSIS

Histomorphology and immunohistochemistry were suggestive of undifferentiated sarcoma favouring extrarenal rhabdoid tumour of soft tissue.

# SUBSEQUENT MANAGEMENT

- In view of persistent headache ,CECT brain was done,
  - revealed right parapharngeal space mass extending to skull base, involving the right jugular  fossa, with thrombus in IJV.

- He was started on ICE based chemotherapy
  - Tolerance of chemotherapy was poor.

- He developed recurrent vomiting and tonic clonic seizures leading to dyselectrolemia (severe hyponatremia) and
- Succumbed to his illness and could not be revived.

# Discussion

- ERRT -Originally described in the kidney
  - reported as “rhabdomyosarcomatous variant of Wilms tumor,”
- Now defined as a distinct clinicopathologic entity
  - with a characteristic clinical course and histopathologic, IHC, and cytogenetics.
- Incidence :- 0.15 per million children <15 years.
- CNS is most frequent site; however,
  - reported in other sites also, like liver, brain, tongue, neck, chest, heart, pelvis, extremities, and several other sites.

- Microscopic analysis shows the presence of a monotonous population of large, round polygonal cells with an abundant eosinophilic cytoplasm, vesicular eccentric nuclei and prominent nucleoli.

- The most striking morphological feature is the deeply, homogeneously acidophilic cytoplasm of the tumour cells, with occasional lateral displacement of the nucleus.

- Immunohistochemically, there is positivity for vimentin and often for keratin and EMA, though generally not for skeletal muscle markers or S-100 protein.
- Defining feature of ERRTs is an aberration (mutation or deletion) of the INI1/SMARCB1 gene located at chromosome 22q11.1. However rare malignant rhabdoid tumours are also SMARCA4 deficient.

- SMARCB1 gene ( INI1, BAF47) is
  - a member of the SWItch/Sucrose Non-Fermentable (SWI/SNF) 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.
    - MRT, AT/RT
    - cribriform neuroepithelial tumor,
    - renal medullary carcinoma, and
    - epithelioid sarcoma.
  - Tumors with variable loss of SMARCB1 expression include
    - subsets of epithelioid malignant peripheral nerve sheath tumor,
    - schwannomas arising in schwannomatosis,
    - subsets of chordomas,
    - myoepithelial carcinomas, and
    - sinonasal carcinomas.
  - Variable and reduced expression of SMARCB1 is characteristic of synovial sarcoma.

# PROGNOSIS AND MANAGEMENT

- Metastases occur in the lungs, liver and lymph nodes.
- The primary tumour must be resected, and the surgical procedure should be followed by postoperative radiotherapy to the primary site and drainage of lymph nodes and chemotherapy.
- Response to therapy is poor and the clinical course extremely aggressive.

THANK YOU