

Case Report

## Atypical Teratoid Rhabdoid Tumor - Two Rare Case Reports

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### ABSTRACT

Atypical teratoid rhabdoid tumors (AT/RT) are rare childhood intracranial tumors. AT/RT represents around 3% of pediatric cancers of the CNS, majority arising in the posterior fossa, below 2 years of age. [1] A germinal mutation showing INI-1 focus is established in some cases of primary renal and CNS neoplasms. Formerly called malignant rhabdoid tumor, it was first described in 1978 as an aggressive variant of Wilms' tumor arising from the kidney. [1-3]

We present two cases of ATRT in 5 month old male baby having Cerebello-Pontine angle tumor and second case is a 4 year old female child having frontal & parasylvian tumor.

**Key words:** Teratoid rhabdoid tumor, Central Nervous system neoplasms, posterior fossa, pediatric.

### CASE REPORTS

A five month old male baby brought by parents with complaints of squint and increased in head circumference, continuous crying and irritability and reluctance to accept feeds.

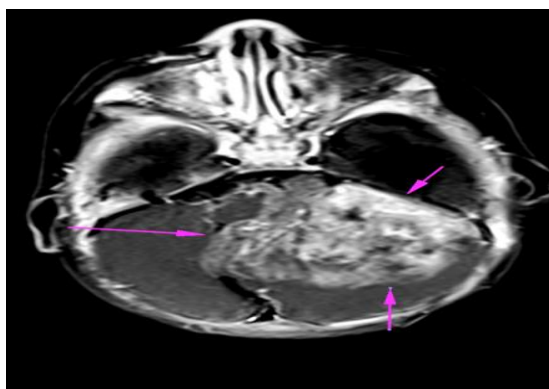


Fig.1 MRI showing mass measuring 5.5x4.7x3.4cm in the posterior fossa.

MRI Findings revealed a large, lobulated, infiltrating mass measuring 5.5x4.7x3.4 cm in size in the posterior fossa epicentered on left side in the region of

cerebello-pontine angle and cerebello-medullary cisterns with extensions. (Fig.1)

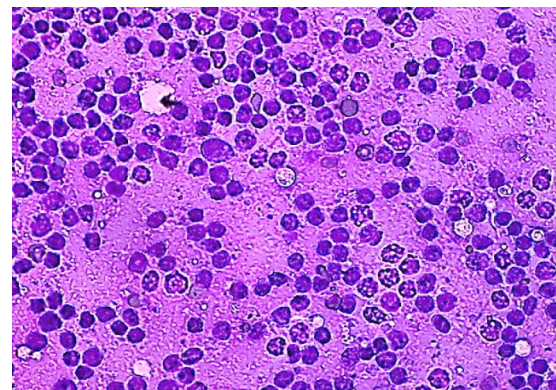


Fig. 2 Photomicrograph of CSF cytology showing presence of malignant round cells.

The patient was operated for right ventricular peritoneal shunt. CSF sent for cytology revealed presence of round blue cell tumor in sheets with the possibility of medulloblastoma. After 2 days the patient underwent left paramedian suboccipital craniotomy with total excision of cerebello-pontine angle intra-axial tumor.

CSF cytology was positive in both the patients showing presence of malignant round cell tumor. (Fig. 2)

The second case is a 4 year old female child having frontal & parasylvian tumor and presented with headache, vomiting & 2-3 episodes of febrile seizures.

The MRI shows a large, oval, intra-axial space occupying lesion measuring 5.8x5.9x7.0 cm in right posterior-lateral frontal lobe adjacent to sylvian fissure with involvement & compressive of anterior portion at basal ganglion. (Fig.3)

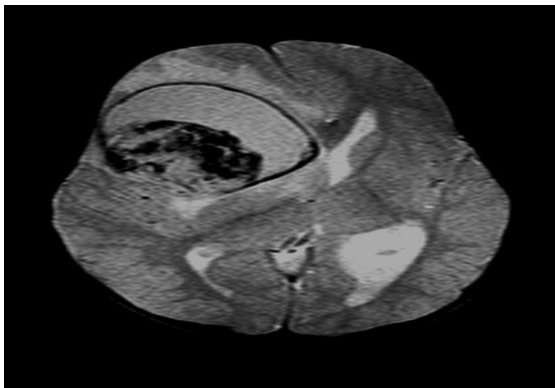


Fig.3 MRI showing mass measuring 7x5.9x5.8cm in right posterior-lateral frontal lobe.

Histological evaluation of both the patients was done, and shows a tumor composed of neoplastic round cell arranged in sheets, small clusters. The neoplastic cells are large, round to oval having large round to oval vesicular nuclei and clumped chromatin and scanty amount of eosinophilic cytoplasm. Pleomorphism & mitotic activity is noted. Necrosis is seen. The intervening stroma is very scanty and shows areas of hemorrhages along with polymorphonuclear & mononuclear cell infiltrate.



Fig.4 Gross image.

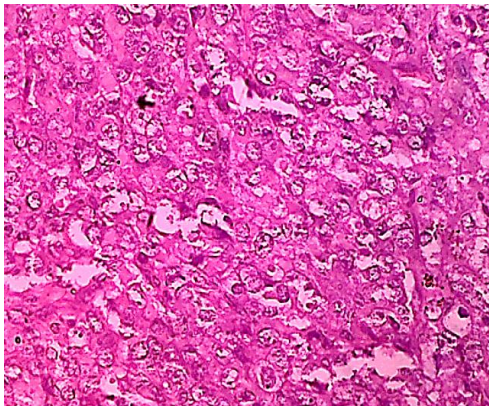


Figure: 5a

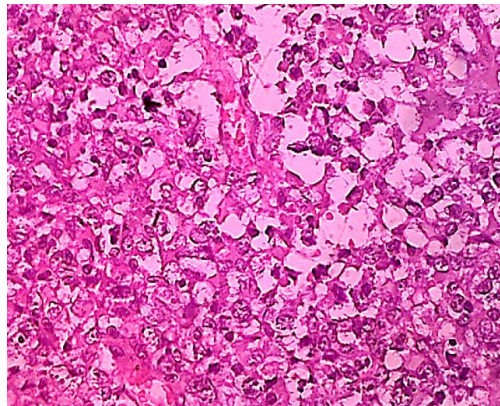


Figure: 5b

Figure: 5 H & E 40X Large pleomorphic neoplastic cells a) Having vesicular nuclei with prominent nucleoli b) Along with rhabdoid cells.

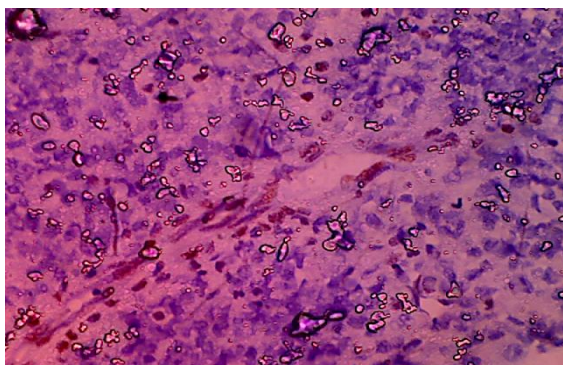


Fig. 6 Figure showing loss of INI 1 protein.

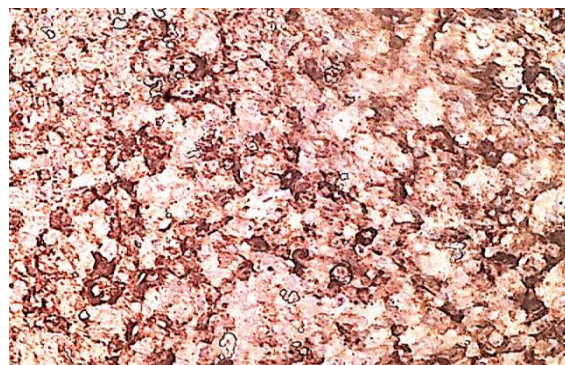


Fig. 7 Figure showing positivity for EMA.

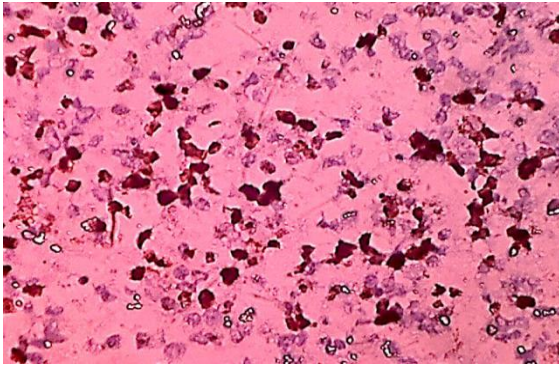


Fig. 8 Figure showing positivity for Mib-1.

## DISCUSSION

AT/RT is a highly malignant central nervous system (CNS) neoplasm that most often occurs in children younger than 2 years of age. [1,2] The tumor is markedly aggressive, with a median survival time of less than 1 year. [1]

Although imaging may suggest AT/RT, diagnosis is based on immunochemistry findings and light microscopy and can be further supported by genetic analysis. Histologically, AT/RT is characterized by the presence of large rhabdoid cells, areas of tumor that resemble PNET, and malignant epithelial and mesenchymal components that lack divergent tissue - differentiation characteristics of malignant teratomas. Immunohistochemically important markers are EMA, vimentin, and smooth-muscle antigen. [4-7]

AT/RT has been misdiagnosed in the past as PNET because of frequent overlapping histologic features. [1-5] AT/RTs can now be distinguished from PNET and other tumors by using specific immunohistochemical markers and by detection of deletions and/or mutations involving the hSNF5/INI1 tumor-suppressor gene in chromosome band 22q11.2. [1-9] These tumors have aggressive growth with high potential for dissemination within the central nervous system (CNS). [1-12] Confirmation of the diagnosis of AT/RT is important because the tumors typically have a poor prognosis that is worse than that of PNET / medulloblastoma (MB), necessitating intensive therapy that differs

markedly from the treatment for PNET/MB. [1,3,8,10-12]

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