

# Case Report: Supratentorial Atypical Teratoid / Rhabdoid Tumor (ATRT) in a Six-Year-Old Child

Raghavendra H<sup>1</sup>, Varsha K S<sup>2</sup>, L N Gupta<sup>3</sup>, Meenakshi<sup>4</sup>

<sup>1</sup>Department of Neurosurgery, ESIC Medical college and Hospital Hyderabad.

<sup>2</sup>Department of Anaesthesia, ESIC Medical college and Hospital Hyderabad.

<sup>3</sup>Department of Neurosurgery, PGIMER & Dr RML Hospital New Delhi.

<sup>4</sup>Department of Pathology, PGIMER & Dr RML Hospital New Delhi

**Abstract:** Atypical teratoid rhabdoid tumor (ATRT) is a rare and highly aggressive malignant tumor of infancy and childhood with fatal outcome. Common site is kidney, but it can also occur in the liver, thymus and the CNS. Primary central nervous system tumor atypical rhabdoid /teratoid tumor is a rare and highly malignant tumor that tends to occur in infancy and early childhood. Most tumors (approximately 2/3<sup>rd</sup>) arise in the posterior fossa. The prognosis of the disease is generally unfavorable. This is a case report of ATRT in an atypical site in a six years old girl.

**Keywords:** ATRT, Supratentorial.

## 1. Introduction

Primary CNS atypical teratoid/ rhabdoid tumor (ATRT) is a rare and highly aggressive malignant tumor comprising 2-3% with an age predilection mainly in less than 2 years. It has also been reported to occur in adults and older children.<sup>1</sup> The supratentorial compartment is less frequently the primary site for this neoplasm, but occasional cases are also arising in the pineal region.<sup>2-5</sup> The histological hallmark of ATRT is the presence of rhabdoid cells.<sup>4</sup> Disseminated forms occur in 20 to 30% cases and generally follow a dismal course with a median time to death is first a few months after diagnosis.<sup>5</sup> We report an unusual case of supratentorial ATRT in six years female child with better outcome.

## 2. Case Report

A previously healthy six years female child who presented with recurrent episode of seizures since one year along with progressive blurring of vision and left hemiparesis since one month. Child was afebrile with intact higher mental function, bilateral no perception of light with primary optic atrophy and left hemiparesis. CT -scan brain showed a large well defined lobulated solid mass in right parieto-occipital region with amorphous calcification (Figure 1) and MRI brain shows large supratentorial solid cystic mass with significant mass effect. Solid component shows intense enhancement while peripheral cystic area show peripheral ring enhancement (Figure 2). MRI spine screening doesn't reveal any spinal metastasis. MRI findings were suggestive of PNET. Child underwent craniotomy and excision of tumor. Tumor was greyish, friable, vascular, diffusely infiltrating brain with areas of calcification. Post operatively child recovered well with persistent vision impairment (Figure 3). Histopathological findings showed multiple friable brownish greyish tissue bits aggregating to 30 cc. On microscopy showed diffuse highly cellular tumor cells arranged in sheets and papillary pattern which are

infiltrating surrounding normal brain parenchyma (Figure 4). Papilla are lined by tumor cells and separated by hyalinized stroma showing vascular proliferation (Figure 5). Tumor cells were displaying typical Rhabdoid Morphology (Figure 6). IHC was performed, the cells were positive for GFAP (Figure 7) and Vimentin (Figure 8), focal positive for EMA (Figure 9) and synaptophysin (Figure 10), negative for S100b, Desmin, SMA. MIB labeling index was 10-11%. Hence reported as atypical teratoid Rhabdoid tumor (ATRT) (WHO grade IV). Post-operative CSF analysis was negative for malignant cells. Post operatively child received both adjuvant Radiotherapy – 60G/30#/6 Wks. and 6 cycles of Carboplatin & Etoposide. Karnofsky performance score (KPS) at one year was 50.

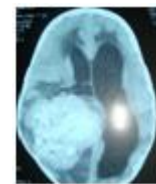


Figure 1: CT-scan showing large mass with calcification

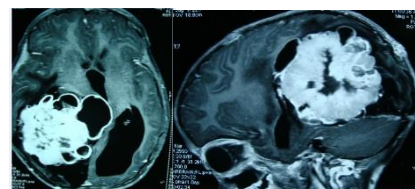


Figure 2: MRI – scan showing large solid cystic lesion with enhancement

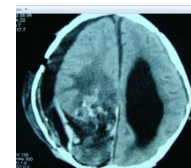
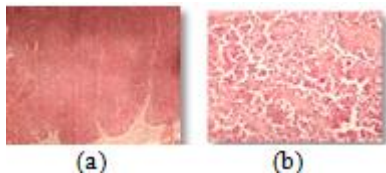
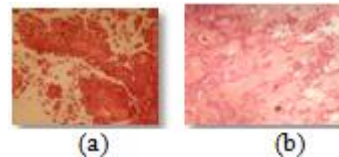


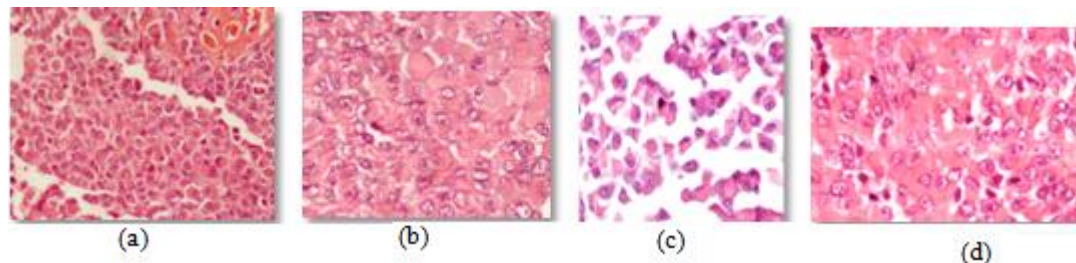
Figure 3: CT-scan post-operative day -1



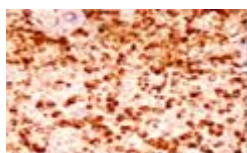
**Figure 4:** Highly cellular tumor arranged in sheets and papillary pattern



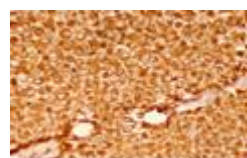
**Figure 5:** Papillae lined by tumor cells and separated by a hyalinized stroma showing vascular proliferation



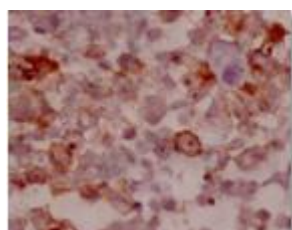
**Figure 6:** Tumor cells displaying the typical rhabdoid morphology: large cells with well-defined cell borders, abundant dense eosinophilic cytoplasm, vesicular nuclei with prominent nucleoli and eosinophilic globular inclusions



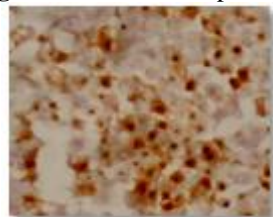
**Figure 7:** GFAP positive



**Figure 8:** Vimentin positive



**Figure 9 :** EMA focal positive



**Figure 10:** Synaptophysin focal positive

### 3. Discussion

ATRT (Grade IV) is a rare and highly aggressive malignant tumor affecting infants and young children.<sup>1,2</sup> They comprises 2% to 3% tumor in children in less than 18 yrs., but majority of patients are younger than 3 years. It frequently affects the posterior fossa. Metastases occur early through the CSF. Other sites affected include the cerebral hemisphere, the pineal region and the spine.<sup>3-5</sup> This is a case report of supratentorial ATRT in a six years old child. Radiological (MRI) findings of PNET/ATRT are similar

hence PNET is the differential diagnosis. ATRT is misdiagnosing as PNET because 70% of ATRT contain fields indistinct from classis PNET.<sup>2,6</sup> The histological features can vary and can resemble medulloblastoma/PNET with small primitive looking neuronal cells with anaplasia. Rosette form can also be seen. Therefore, it is therefore important to have differential diagnosis of ATRT in mind especially in younger children.<sup>7</sup> In present case tumor shared the microscopic features of PNET/ ATRT. On IHC, ATRT stain positive for Vimentin and focally or weakly positive for EMA, GFAP, and SMA. Proliferative activity is high as the labeling index with Ki- 67/MIB-1. Present case showed strong positivity for Vimentin and GFAP with MIB -1 is 10-11% positive. S-100, Desmin, SMA were negative. Thus, confirmed the diagnosis of Atypical teratoid/ rhabdoid tumor (ATRT) (WHO grade IV).

ATRT show monosomy for chq 22 and tumor suppressor gene chq 22 .911.2 which helps to distinguish from other PNET including medulloblastom.<sup>2,7</sup> INI 1 gene is implicated in the intra cranial ATRT both by somatic and by germ line mutation. ATRT lack the INI-1 gene product.<sup>8,9</sup>

These tumors are highly aggressive and have malignant local infiltration making total excision infeasible. Prognosis is dismal with median survival of 6 months only. When associated with CSF dissemination survival in just 2.5 months. Present case shows one year survival with KPS of 50.

Because of its rarity and rapid course, there has been no concern as to the optimal treatment of this tumour. The optimal treatment for ATRT remains unclear, surgery mainly for cytoreduction along with adjuvant chemoradiotherapy.<sup>10</sup> Chemotherapy also has part to play in treatment of this tumor, especially in children younger than 3 years in whom radiation therapy to be delayed due its deleterious effect on the developing CNS. Children who receive surgery without adjuvant therapy can die within a month. Recurrent ATRT in less than 3 years of years of age are chemo resistant. Debulking followed by chemotherapy with single agent cyclophosphamide, Ifosphamide with or without radiotherapy considering the age of patient.<sup>10</sup>

#### 4. Conclusion

Though atypical teratoid/ rhabdoid tumor (ATRT) is rare in supratentorial location which is more commonly seen in children less than 3 years, it should be always kept in mind older children, due to its fatal clinical outcome and aggressive behavior, MRI, histological features and IHC play a crucial role in the diagnosis. Older children have better prognosis than younger counterparts.

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