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Case Report *Volume 6 Issue 1*

An Interesting Rare Case Report of CNS Atypical Teratoid Rhabdoid Tumour in 8-Year-Old Male

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Abstract

Atypical teratoid rhabdoid tumor (AT/RT) is a rare and aggressive malignancy predominantly diagnosed in childhood. Although it primarily arises in the brain, AT/RT can occur anywhere within the central nervous system (CNS), including the spinal cord. Approximately 60% of cases are in the posterior cranial fossa, particularly the cerebellum. This report presents the case of an 8-year-old male with no prior medical history diagnosed with this rare tumour.

Keywords: Atypical Teratoid; Rhabdoid Tumor; Central Nervous System; Cerebellum; Tumour

Abbreviations

CNS: Central Nervous System; SMA: Smooth Muscle Actin; EMA: Epithelial Membrane Antigen; FISH: Fluorescent In Situ Hybridization; PNET: Primitive Neuroectodermal Tumours.

Introduction

Atypical teratoid/rhabdoid tumour (AT/RT) is a highly malignant CNS embryonal tumor characterized by poorly differentiated elements, often featuring rhabdoid cells. It is typically associated with the inactivation of the SMARCB1 (INI1) gene or, in rare cases, SMARCA4 (BRG1). AT/RT is most commonly diagnosed in infants and young children under the age of 3, with a mean age of approximately 2 years. While rare in children over 6 years old, occasional cases have been reported. The tumor exhibits a male predominance and is frequently found in the cerebral hemispheres, followed by the cerebellum, cerebellopontine angle, and brainstem, with spinal involvement being exceedingly rare. Histologically, AT/RT is composed of rhabdoid cells, often accompanied by areas resembling primitive neuroectodermal tumour, epithelial tissue, neoplastic mesenchyme, and neuronal or glial differentiation [1,2].

In the United States, AT/RT accounts for approximately 3% of pediatric CNS tumours, with an incidence of three cases per 1,000,000 children annually. CNS tumours represent about 17% of all pediatric cancers, making them the most common solid tumours in children. The overall survival rate for CNS tumours is approximately 60% [3-5].

Case Report

An 8-year-old male presented to the outpatient department with a two-week history of headaches. The patient described the pain as holocranial, dull, and aching, with mild to moderate intensity. The headache was non-radiating, showed no diurnal variation, and had no identifiable aggravating factors. It was partially relieved by oral NSAIDs. The patient

had no other medical complaints, and the neurological examination revealed no significant abnormalities [6,7].

Brain MRI revealed a heterogeneously enhancing, well-defined intra-axial lesion measuring 61mm x 54mm x 49mm in the left inferior temporal lobe and parahippocampal gyrus [8]. The lesion exhibited a broad base along the tentorial surface, appearing hypointense on T1-weighted images, hyperintense on T2-weighted and FLAIR sequences, and showing blooming on GRE with mild diffusion restriction on DWI. The tumour indented the periventricular white matter on the left side, with a CSF cleft visible along the lesion. Mild perilesional edema was observed, along with areas suggestive of necrosis, calcification, or hemorrhage. The mass effect resulted in effacement of the underlying cortical sulci and a 3mm midline shift to the right (Figures 1 & 2).

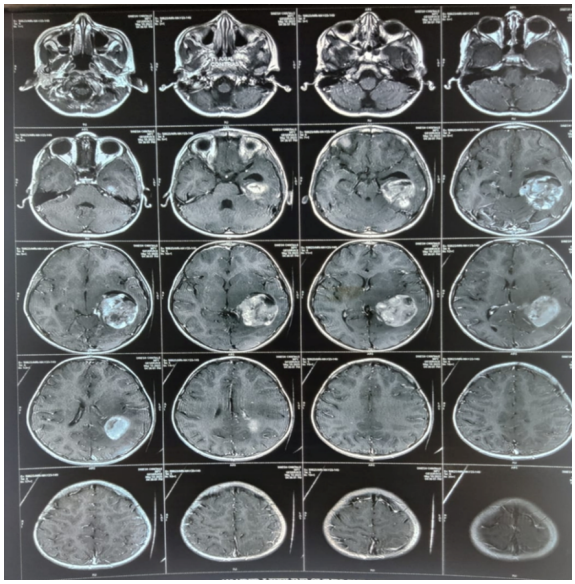

Figure 1: Brain MRI.

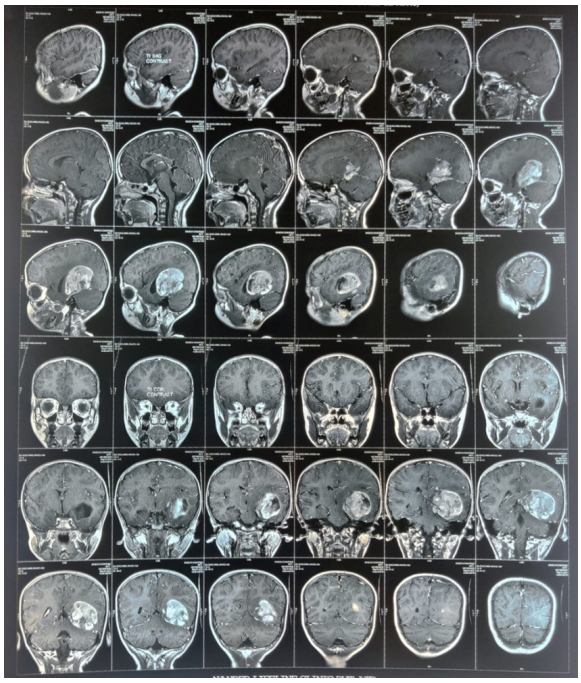

Figure 2: The patient underwent a right parietotemporal craniotomy for tumour excision, followed by radiation and chemotherapy.

Discussion

AT/RT was first described as a distinct CNS entity in 1985. Rorke LB, et al. [1] later characterized it as an "atypical teratoid/rhabdoid tumour" due to its heterogeneous composition of rhabdoid, primitive neuroepithelial, epithelial, and mesenchymal elements. Notably, AT/RT shares similarities with malignant rhabdoid tumours of the kidney and exhibits a male predilection.

Imaging Findings

On imaging, AT/RT typically appears as a hyperdense lesion on unenhanced CT scans, with heterogeneous contrast enhancement. MRI findings include hypointensity on T1-weighted images and gadolinium enhancement, consistent with the findings in this case [9].

Gross Pathology

Grossly, AT/RTs are soft, pinkish-red masses that are partially demarcated from surrounding brain tissue. They often contain areas of hemorrhage and necrosis, as observed in this patient [10].

Immunohistochemistry and Genetics

AT/RTs are polyphenotypic tumours that commonly express epithelial membrane antigen (EMA), vimentin, and smooth muscle actin (SMA). Other markers, such as GFAP, neurofilaments, S-100, synaptophysin, and keratin, may also be detected depending on the tumour's cellular composition. Loss of INI1 nuclear protein expression, a hallmark of AT/RT, is observed in most cases [11].

Cytogenetics

Cytogenetic studies, particularly fluorescent in situ hybridization (FISH), can help differentiate AT/RT from other CNS tumours, such as medulloblastoma or primitive neuroectodermal tumours (PNET).

AT/RTs frequently exhibit deletions in chromosome 22q11.2, while medulloblastomas often show deletions in chromosome 17p. The presence of hSNF5/INI1 gene mutations is a key diagnostic feature of AT/RT [12].

Management

Surgery

Surgical resection is critical for diagnosis but is rarely curative due to the tumour's aggressive nature and challenging locations, such as the cerebellopontine angle. Disseminated disease is present in approximately one-third of patients at diagnosis [13].

Chemotherapy

Chemotherapy alone is rarely curative, though transient responses are observed in about 50% of cases. Various agents, including cisplatin, carboplatin, cyclophosphamide, vincristine, and etoposide, have been used. Intrathecal chemotherapy, delivered via an Ommaya reservoir or lumbar puncture, is also employed to bypass the blood-brain barrier [14].

Radiation Therapy

Radiation therapy is often deferred in children under 3 years due to long-term complications. However, its upfront use is increasingly considered in AT/RT due to the poor prognosis. External beam radiotherapy (EBRT) is the preferred modality [15].

Prognosis

The prognosis for AT/RT remains poor, with a two-year survival rate of less than 20% and an average postoperative survival of 11 months. However, recent multimodal treatment protocols have shown improved outcomes, with some studies reporting a 70% survival rate at 2–3 years.

Retrospective analyses from institutions like St. Jude Children's Hospital and Cleveland Children's Hospital highlight the importance of age and treatment intensity in survival outcomes.

Conclusion

AT/RT is a rare and aggressive CNS tumour with a challenging prognosis. Early diagnosis, multimodal treatment, and advances in genetic profiling are essential for improving outcomes.

This case underscores the importance of considering AT/RT in older children presenting with atypical CNS lesions with recent advances in molecular and genetic diagnosis, we have been able to identify several genetic markers for this cancer which have laid the foundation for further use of gene therapy and more targeted molecular therapies.

Early diagnosis and surgery however remains the mainstay of treatment with adjuvant radiotherapy and chemotherapy. The tumour is highly vascular with areas of necrosis within, which makes it technically difficult to remove.

In view of this there is a shift from the concept of Gross total resection to a maximal safe resection to prevent neuro deficits. Despite the latest available modalities of radio and chemo therapy the overall outcome remains poor with a high rate of mortality and high chance of recurrence.

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