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CASE REPORT

ATYPICAL TERATOID/RHABDOID TUMOR: A RARE CASE REPORT

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ABSTRACT

Atypical Teratoid/Rhabdoid Tumors (AT/RTs) is rare, highly malignant embryonal tumor of central nervous system that predominantly occurs in infancy and young children. The majority of tumors arise (approximately two-third) in the posterior fossa. The diagnosis of AT/RTs relies predominantly on morphologic and immunohistochemical criteria. The most common differential diagnosis are Primitive Neuroectodermal Tumor and malignant glioma. Herein we present a case of 5 year old child who initially presented with headache and vomiting.

Key words: *Atypical, Teratiod, Tumor*

INTRODUCTION

Atypical teratoid/rhabdoid tumor (AT/RT) is a highly aggressive neoplasm. They were termed "AT/RT" because they contain nests or sheets of rhabdoid tumor cells as well as varying proportions of primitive neuroectodermal tumor cells, mesenchymal spindle-shaped tumor cells, and/or epithelial-type tumor cells 1. AT/RT has been misdiagnosed in the past as PNET because of frequent overlapping histologic features 2. 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 22q11.23-5. The large rhabdoid cells seen in AT/RT were described first in association with a unique pediatric kidney neoplasm termed rhabdoid tumor of infancy6-8. The rhabdoid phenotype subsequently was noted in extrarenal sites as well, including the CNS. Rorke et al.9 at The Children's Hospital of Philadelphia in 1987, coined the term "AT/RT" to describe these biologically unique neoplasms with aggressive behavior and characteristic morphologic features 9-10.

Here in we report a case of AT/RT occurring in posterior fossa in a 5 year old male child.

CASE REPORT

A previously healthy 5 year old boy was evaluated for headache and multiple episodes of vomiting for

last one month duration. Headache was intermittent associated with non- projectile vomiting. MRI of head showed complex mass in left parieto-occipital lobe. CT head showed 4x7 cm heterogeneous solid intraventricular mass within the occipital horn of left lateral ventricle. There is associated dilatation of the left lateral ventricle and mild dilatation to the right lateral ventricle. There is a density calcified focus within it. Other routine investigations were normal. The patient underwent left Mittre's flap craniotomy with excision of tumor and placement of External Ventricular Drain. Histopathology of the excised tumor showed atypical teratoid/rhabdoid

tumor who Grade IV. Immunohistochemical study tumor was positive for vimentin. Patient is now started on Craniospinal radiation.

MRI Images:

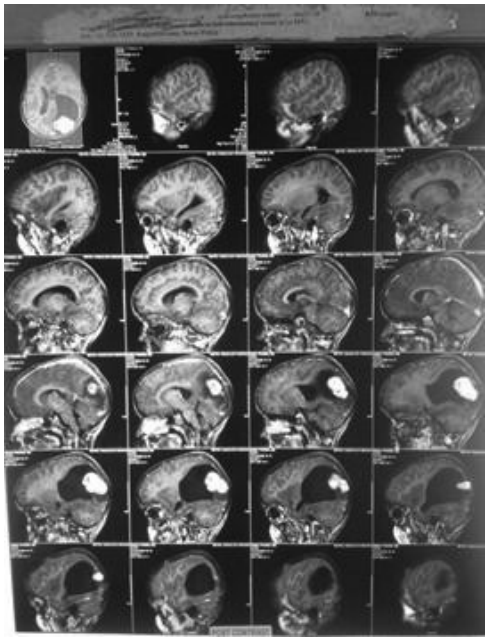
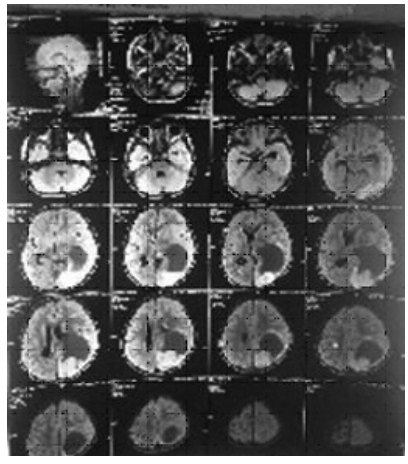


Fig: MRI and CT scan of brain showing tumor in the posterior fossa

DISCUSSION

AT/RTs of the CNS are aggressive childhood neoplasms that are located most commonly in the posterior intracranial fossa11. AT/RT is a rare primary malignant tumor of the CNS with an incidence of 2-3%.Two thirds of these cases occur in the pediatric age group with a slight male preponderance6-8. Imaging studies, particularly the use of magnetic resonance imaging, are useful initial diagnostic modalities. Most of the lesions are bulky, contrast enhancing with hemorrhage and necrosis12. The histopathologic spectrum of AT/RT is broad, ranging from predominantly "small cell" with primitive morphology to tumors with large rhabdoid cells. In addition, some AT/RTs may have mesenchymal and epithelial components. Because of this morphologic variability,AT/RTsoftenhavebeenmisclassified2,6,11 AT/RT can now be distinguished from PNET/MB by using specific immunohistochemical markers (EMA,



vimentin)and mutation analysis. Confirmation of the diagnosis of AT/RT is important because these tumors typically have a poorer prognosis than that in PNET/

MB, necessitating new intensified therapies. The overall 1-year and 5-year survival probabilities were 71% and 28%, respectively1.

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