Research Paper

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Intracranial Atypical Teratoid/ Rhabdoid Tumor in a child: A Rare Case Report

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INTRODUCTION

Rhabdoid tumour was originally described in 1978 by Beckwith et al.[1] as a highly malignant paediatric tumour of the kidney, and Rorke et al.[2] named primary central nervous system malignant rhabdoid tumour as 'atypical teratoid/rhabdoid tumor (ATT/ RhT). ATRTs were identified as separate entity as recognized by their addition in WHO classification of tumors in 1993[7]..These tumors resemble rhabdomyosarcomas, but their immunohistochemical and ultrastructural features allow a clear distinction between the two types of tumors. This biologically aggressive CNS neoplasm occurs both in posterior fossa and in supratentorial compartment. The tendency to arise in the cerebropontine angle with invasion of surrounding structures is a distinct feature of ATT/RhT. Recently, some authors have reported a small number of cases of extrarenal rhabdoid tumors. Primary central nervous system (CNS) malignant rhabdoid tumors are rare. Since 1985, 85 cases of primary CNS malignant rhabdoid tumor have been documented[2,3,4,5,6]. Intracranial ATT/RhT is a disease of infancy and childhood, usually occurring during the first decade of life[2]. The most common biologically malignant CNS tumor in this period is a primitive neuroectodermal tumor-medulloblastoma (PNET-MB). ATT/ RhT has been frequently misdiagnosed as a PNET-MB because 70% of ATT/RhTs contain histological fields indistinguishable from classic PNET-MB. However, microscopic features in routine and special studies allow the differential diagnosis of PNET-MB and ATT/RhT[1,2,3,4,5,6]. We report a case of a primary CNS ATT/RhT arising from the posterior cranial fossa in a child. The clinical course of the patient is described with a special emphasis on the differential diagnosis. Primary CNS ATRTwas recognised as a separate entity and added to the World Health Organisation (WHO) classification of tumors in 1993 as a grade IV embryonal tumor[9,10]. Immunohistochemistry plays a role in confirming the diagnosis with loss of INI-1 staining in neoplastic cells[11].

CASE REPORT

We present a case of 2 year female who came to the surgical OPD with chief complaints of left sided weakness of body since 4 month. General examination revealed no abnormality. Haematological and biochemical results were within normal limit. Magnetic Resonance Imaging showed a large lobulated intra axial lesion in the right temporo parietal lobe measuring 5x4cm with calcification in posterior fossa. Excision of the suspected mass was done and sent for histopathological examination. Grossly the mass was 5x4 cm, grayish, friable and highly vascular.

Microscopically the tumor showed aggregation of large polygonal cells , with vesicular nucleus, prominent nucleoli and moderate amount of eosinophilic or pale cytoplasm, referred to as rhabdoid cells. Some of the cells showed intracytoplasmic eosinophilic hyaline material displacing the nucleus to the periphery thereby giving the classical eccentric rhabdoid appearance.

Immunohistochemical study revealed focal positivity for vimentin, epithelial membrane antigen(EMA). They were totally negative for glial fibrillary acidic protein (GFAP). Based on histological findings along with confirmation with immunohistochemical markers, final diagnosis of Atypical tertatoid/ Rhabdoid tumor was given.

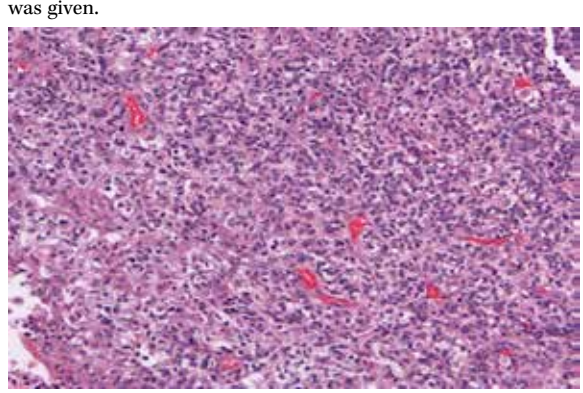

Fig:1 H&E Sections show atypical highly pleomorphic cells arranged in sheets as well as in small clusters.

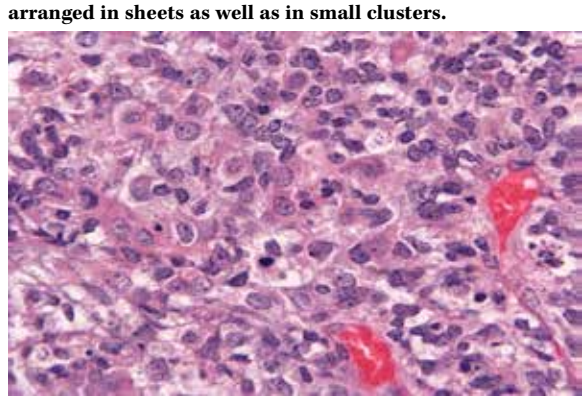

Fig:2 H&E Sections show presence of highly pleomorphic rhabdoid cells with variable amount of eosinophilic cytoplasm with prominent nucleoli, with few showing hyperchromatic nuclei.

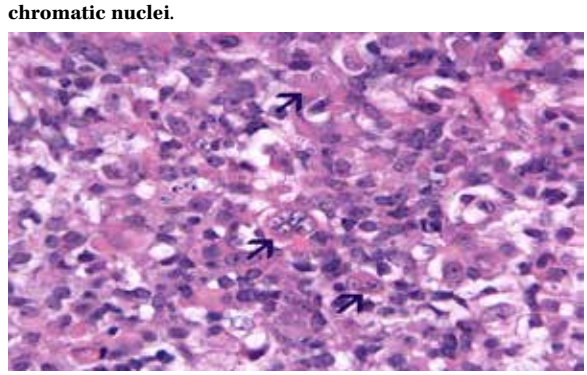

Fig:3 H&E Sections show presence of highly pleomorphic multinucleated cells (arrows) with variable amount of eosinophilic cytoplasm with prominent nucleoli.

Discussion

ATRT remains a significant challenge in pediatric neurooncology and difficult to classify due to overlapping morphologic and immunohistochemical findings. AT/RT is a tumor of infancy and childhood, and very rare in adults[5,6,7]. The mean age of the patients is 2.9 yr and three-quarters of them are 3 yr or younger at the time of diagnosis with a male predominance[8]. Childhood PNET-MB, tends to appear between 3 and 5 yr of age. Eighty percent of classic PNET-MBs arise in the cerebellum, but the AT/RTs develop on that site in only slightly over half [3-6, 8]. The cerebellopontine angle appears to be a common location for AT/RT; 15% in series of Rorke et al[6]. AT/ RT is a highly malignant neoplasm involving leptomeningeal dissemination in 10-30% of patients[3-6,10].

Clinical presentation depends on the age of onset and the location of the tumor. Children younger than 3 yr of age usually present with nonspecific symptoms and signs, such as vomiting, lethargy, irritability, loss of weight, macrocephaly, and failure to thrive. Older patients commonly present with increased intracranial pressure or localizing signs. Cranial nerve palsies, headache, and hemiplegia are common[2-4,6]. The characteristic findings of ATRT are heterogenous density on pre contrast CT and homogenous enhancement. Differential diagnosis between ATRT and PNET-MB is difficult by radiological findings but is easier by histopathological findings. Light microscopic examination of ATRT reveals a diffuse growth patternwith predominantly polygonal cells arranged in a focally trabecular and alveolar pattern. The individual cells have vesicular nuclei with prominent nucleoli, few cells showing hyaline cytoplasmic inclusion.

In summary, we report a case of 2 year female who presented with chief complaints of left sided weakness of body since 4 month. Magnetic Resonance Imaging indicated PNET. Histopathological findings was consistent with ATRT. CNS ATRT is very rare and prognosis is invariably poor and hence merits reporting.

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