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Rare presentation of atypical Teratoid / Rhabdoid tumor : Case report and review of literature

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ABSTRACT

Case report: An 18 year old female presented with recurrent generalized tonic clonic seizures of four years duration. After the first episode she was evaluated at a local hospital and was detected to have a right parieto-occipital lesion and was advised surgery but she defaulted from treatment. She presented with a 30 day history of headache and vomiting and on examination was alert with stable vital signs. She had left homonymous hemianopia and fundus examination revealed papilledema. She did not have any other cranial nerve or focal neurological deficits. Her systemic examination was unremarkable. Computerized tomogram of brain showed a large heterogeneous cortical based lesion in the right parieto-occipital lobe with enhancing hyper dense irregular wall and central hypodensity suggestive of necrosis. Small area of calcification was seen in the inner wall of the lesion. There was no perilesional oedema. (Figure 1A, B). Her magnetic resonance imaging which was done earlier showed a right parieto-occipital lesion which was isodense on T1 weighted sequences, hyperintense on T2 weighted and FLAIR sequences with irregular contrast enhancement. There was no perilesional edema. (Figure 1 C, D)

INTRODUCTION

A right parietooccipital craniotomy and total decompression was performed. The tumor was vascular and amenable to suction with varying consistency. It had good plane of demarcation from surrounding brain. Histopathological examination showed a highly cellular as well as infiltrating neoplasm composed by sheets of neoplastic glial cells and these cells are supported on a fibrillary matrix. At several microscopic foci these cells have an eccentric nuclei and abundant eosinophilic cytoplasm resembling 'rhabdoid' cells. There was characteristic spread of the neoplastic cells along 'Virchow-Robbin' spaces with discrete zones of necrosis and nodular masses of endothelial proliferation. There were hypodense apoptotic cells and few mitotic figures. (Figure 1 E, F). The 'rhabdoid cells' gave positive immunostaining for vimentin and the MIB index was more than 8 percent in different microscopic fields. Patient did not have any post-operative deficits and underwent thirty three cycles of radiotherapy. Six months after surgery, she is doing well functionally with no evidence of recurrence in the post operative images.

Discussion: Atypical teratoid/rhabdoid tumors (AT/RTs) are rare malignant intracranial tumors, predominantly occurring in posterior fossa of pediatric population. They form representing 6.7% of CNS tumors in children younger than 2 years[1]. Malignant rhabdoid tumors are reported to occur in many locations in the body, though the kidney and

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CNS are the most common primary sites. Intra cranial AT/RTs have a 3:1 male predominance and are mostly infratentorial (38%–65%) and 27%–62% occur in the supratentorial locations especially on the left side[2]. Rorke et al reported AT/RT affecting the nervous system in 1985 and coined the term AT/RT as the tumor was a combination of rhabdoid, primitive neuroepithelial, epithelial and mesenchymal components[3,4].

The pathogenesis of AT/RTs is not clearly understood. Recent hypothesis supports the presence of pluripotent cell with the ability to develop into epithelial and mesenchymal cells[5]. Transformation of a stable brain neoplasm to aggressive AT/RT due to acquisition of HIV-integrase interactor 1 (INI1) gene mutation with a characteristic rhabdoid morphologic change has been suggested by Allen et al.[5] Our patient also had clinical and imaging evidence of a tumor which later showed malignant change after four years of diagnosis.

Most cases present with short history of raised intracranial pressure and focal deficits. Seizures can also be a presenting symptom.[4,5]. Imaging findings on brain computed tomogram (CT) is that of an iso- or mild hyperdense lesion in relation to gray matter which shows contrast enhancement, and may have associated parenchymal edema, calcification or hemorrhage. Multiple cystic or necrotic foci are also seen.[1] Lesions are usually of large size at presentation. Tumor may be located within or extending into the ventricles. Destruction of the overlying skull and extracranial extension have been described[2]. On MRI scan, tumor shows mixed signal intensity on T1 and T2-weighted images due to extensive necrosis and intratumoral hemorrhage. The solid component is hypo- or isointense on T1-weighted images and iso- or hyperintense on T2 -weighted images compared to the gray matter with good contrast enhancement.[1,2] Leptomeningeal spread can also be detected by MR imaging and is an indicator of poor prognosis.[2]

Histologically AT/RT is composed of rhabdoid cells or is combined with fields indistinguishable from PNET with neoplastic mesenchymal or epithelial tissue. Rhabdoid cells have abundant eosinophilic cytoplasm with eccentric nuclei, prominent nucleoli and hyaline globular inclusion which are aggregates of intermediate filaments.[5] The rhabdoid cells usually express vimentin and EMA with variable expression of GFAP, S-100 and cytokeratin. INI1/hSNF5 (HIV-integrase interactor 1 (INI1)/human homolog of Saccharomyces Cerevisiae sucrose-nonfermenting 5 (hSNF5), mutation and/or loss of INI1 protein expression is often quoted as a diagnostic tool for detection of AT/RT.[5] The presence of necrosis and high proliferation index (Mib1) indicates the aggressiveness of the tumor.

AT/RT have tendency to spread with leptomeningeal involvement[2]. So, contrast-enhanced MR of the brain and spine should be performed during follow-up as poor prognosis is associated with MR imaging evidence of disseminated leptomeningeal tumors. Surgical decompression followed by radiotherapy and chemotherapy is the treatment of choice.[5] Reported mean survival ranges from 6 to 15 months from diagnosis but longer survival has been reported in adults. The fact that a patient survived 17 years after total tumor resection and adjuvant multimodality therapy indicates the role for these therapeutic modalities in better survival of adult patients[5]. The case presented here is a rare presentation of AT/RT; with a long history, atypical location in an adult patient.

Conclusion : Atypical teratoid/rhabdoid tumor should be considered as a differential diagnosis when a cortical enhancing lesion is being evaluated in an adult. Better survival is expected in adults with multi-modality treatment schedules.

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