

Atypical Teratoid/Rhabdoid Tumor in Adults an Uncommon Entity: A Case Report

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Abstract

Atypical teratoid/rhabdoid tumor in adults is a relatively rare malignant neoplasm. It is characterized by the presence of rhabdoid cells in combination with loss of either the INI1 or BRG1 protein from the tumor cells. We herewith present an adult patient with 4th ventricular lesion, which on histopathology turned to be AT/RT an uncommon finding in adults.

Keywords: *Atypical teratoid/rhabdoid tumor; Adult*

1. Introduction

Atypical teratoid/rhabdoid (AT/RT) tumor is a rare, highly malignant tumor of the central nervous system (CNS), most commonly found in children less than 5 years of age. Although the vast majority of cases are diagnosed in young children, there have been isolated case reports in adults. Since its histological appearance can be confused with other tumors, especially in adults, separating AT/RT from other neoplasms may be difficult. In many instances, a reliable diagnosis is not possible without demonstrating the lack of nuclear INI1 (*SMARCB1*) or *BRG1* (*SMARCA4*) protein expression by immunohistochemical methods or by detection of somatic / germline mutation of the INI1 (*SMARCB1*) or *BRG1* (*SMARCA4*) gene. Final diagnosis is confirmed after immunohistochemical analysis and/or molecular analysis. Immunohistochemical staining show that the tumor cells are positive for vimentin and reacted variably for keratin, epithelial membrane antigen (EMA), synaptophysin, neurofilament protein, CD34, and smooth muscle actin (SMA) and negative for GFAP, S-100, desmin and CD99. In adult examples of AT/RT, the diagnosis requires a high index of suspicion, with early tissue diagnosis and a low threshold for investigation with INI1 immunohistochemistry to differentiate this entity from other morphologically similar tumors. Although

the prognosis is dismal in pediatric population, long term survival is possible in adult AT/RT cases after surgery, adjuvant radiotherapy and chemotherapy.

2. Material and Methods

This 30-year-old patient was admitted in our hospital with complaints of headache and nausea. On examination no gross deficit was seen. Patient underwent MRI brain which revealed contrast enhancing 4th ventricular tumour with mass effect as per FIG. 1. Patient was explained need of surgical excision and all possible risks and benefits involved.

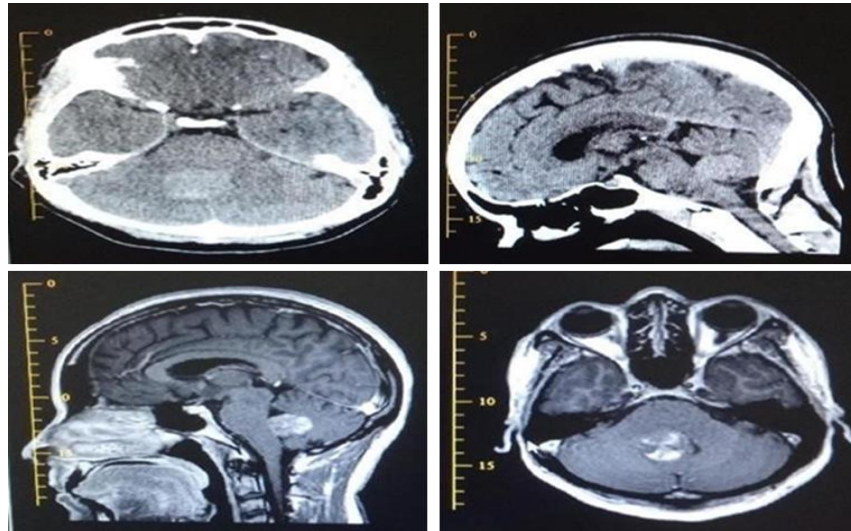


FIG. 1. Pre-operative CT scan and MRI images.

3. Results

Once patient agreed, a sub-occipital craniotomy was done, and total excision of 4th ventricular tumour was performed. Postoperative period was uneventful. Postoperative scan was satisfactory as per FIG. 2. Patient had some gaze palsy owing to brain stem proximity and is recovering gradually. Patient was discharged home with referral for radiation oncology and is on regular follow up in OPD. Histopathology revealed the tumor to be an Atypical teratoid/ rhabdoid tumor, WHO grade IV, with loss of nuclear INI1 (*SMARCB1*) immunostain, an uncommon entity in adults as per FIG. 3 & 4.

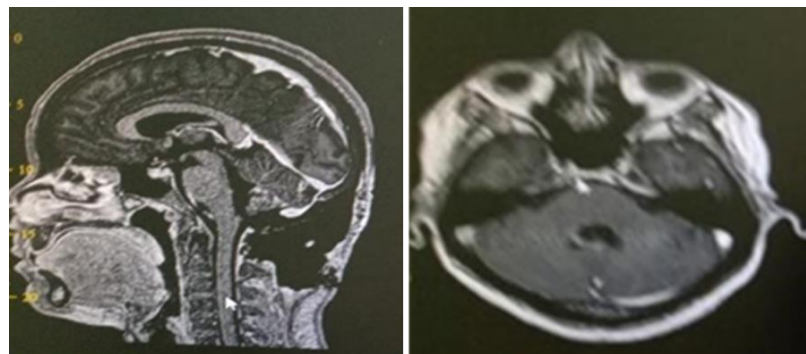


FIG. 2. Post-operative MRI of the patient.

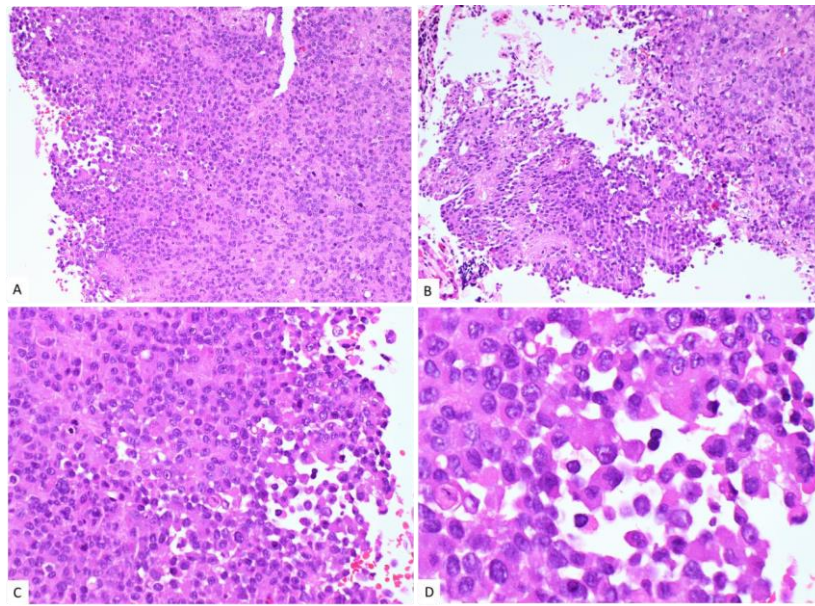


FIG. 3. A, C. Atypical teratoid/rhabdoid tumor composed of poorly differentiated cells intermixed with cells showing rhabdoid features such as eccentric nuclei and abundant eosinophilic cytoplasm (H&E, A: 20× magnification, B: 40× magnification). B. Atypical teratoid/rhabdoid tumor with focal perivascular pseudorosette pattern (H&E, 20× magnification). D. Rhabdoid cells showing eccentric nuclei with vesicular chromatin and prominent nucleoli, and eosinophilic globular cytoplasmic inclusions (H&E, 60× magnification).

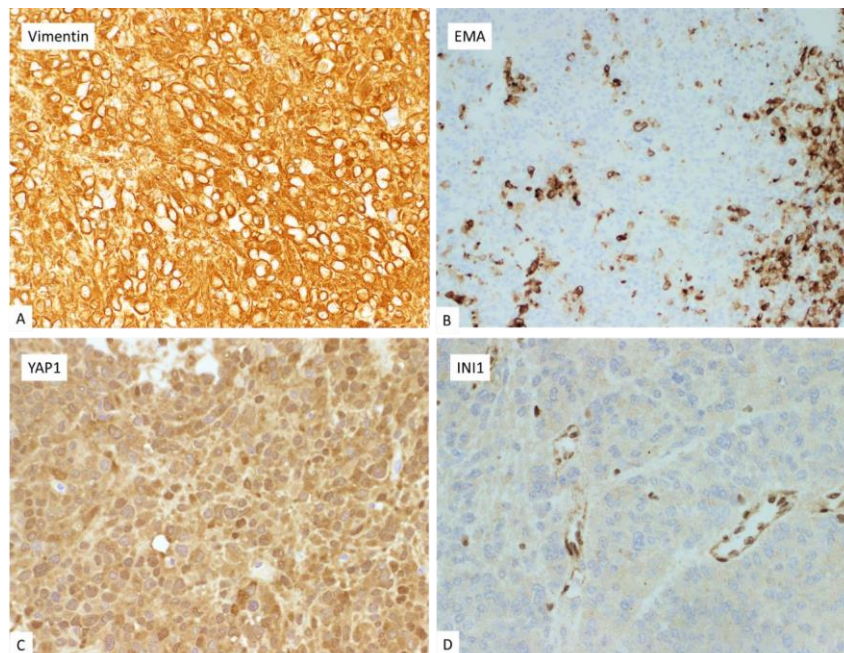


FIG. 4. Atypical teratoid/rhabdoid tumor. A. Strong expression of vimentin immunostain (40× magnification). B. Epithelial membrane antigen (EMA) immunostain shows patchy positive membranous and cytoplasmic expression (40× magnification). C. YAP1 immunostain shows cytoplasmic and nuclear expression (40× magnification). D. Loss of expression of SMARCB1 (INI1) immunostain in nuclei of tumor cells, with retained expression in the intratumoral blood vessels (40× magnification).

4. Discussion

In 2014, Shitara et al reported Atypical teratoid/rhabdoid tumor in sellar turcica in an adult [1]. In 2010, Las Heras reported an Atypical teratoid/rhabdoid tumor of the spine in an adult [2]. In 2007, Zarovnyaya EL et al. reviewed the literature of Atypical teratoid/rhabdoid tumor with ganglioglioma-like differentiation [3]. In 2014, Krishnan C et al. described their two cases of Atypical teratoid/rhabdoid tumor with ganglioglioma-like differentiation and reviewed the literature [4]. In 2017, Dardis C et al. described their two cases of same pathology and discussed their implications for pathophysiology and treatment [5]. In 2016 WHO Classification of Tumors of the Central Nervous System defined AT/RT by alterations of either INI1 protein (*SMARCB1* gene), or rarely, BRG1 protein (*SMARCA4* gene) [6]. Recent studies using multimodal therapy have shown significantly improved survival data. In 2008, the Dana-Farber Cancer Institute in Boston reported two-year overall survival of 53% and event-free survival of 70% [7]. In 2013, the Medical University of Vienna reported five-year overall survival of 100%, and event-free survival of 89% [8].

5. Conclusion

To conclude Atypical teratoid/rhabdoid tumor is an uncommon pathology in adult and our case is one of them. Outcome is better in adults than children and radiotherapy is the mainstay of treatment after surgical excision.

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