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Atypical Teratoid / Rhabdoid Tumor of the Sellar region: Case Report and Review of the Literature

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Abstract

Atypical teratoid rhabdoid tumors (AT/RTs) are rare and aggressive pediatric malignant rhabdoid tumors (MRT) that occur within the brain. The majority of these tumors occur in the cerebellum. Only 45 cases of adults with AT/RT have been reported in the literature to date. We present a case of sellar and suprasellar AT/RT in a 40-year-old female patient with this rare entity. To our knowledge, this is the 14th case of an adult-onset AT/RT in the sellar and suprasellar region.

Key words: Atypical teratoid / rhabdoid tumor, Sellar/suprasellar lesion, Surgery

Introduction

Atypical teratoid rhabdoid tumors (AT/RTs) are rare and aggressive pediatric malignant rhabdoid tumors (MRT) that occur within the brain [1]. It is initially reported in 1987 and subsequently, defined as a distinct CNS neoplasm in 1996. It is added to the World Health Organization (WHO) Brain Tumor Classification in 2007 (grade IV) [2,3]. Histologically, ATRT is known that they are composed of diffuse proliferation of atypical large cells showing eccentrically located nuclei and abundant eosinophilic cytoplasm (rhabdoid features) with prominent nucleoli [4]. AT/RTs are characterized by biallelic loss of SMARCB1. Here, we report a case of AT/RT that originated in sellar and suprasellar region in a 40-year-old female patient with brief review of this aggressive tumor in adult population.

Case report

40-year-old right-handed female not known to have any medical illnesses, presented to the emergency department with a history of severe headache and decrease vision, which was more on the right eye. It was associated with nausea and vomiting for the last few days and photosensitivity as well. Clinical examination revealed decreased vision of the right eye and third nerve palsy of the same side. She denied any history of prolactinoma, constitutional symptoms and family history of pituitary tumors. MRl and CT scan showed sellar and suprasellar lesion that was invading the cavernous sinus bilaterally but more into the right side and protruding through the sella turcica. Preoperative magnetic resonance imaging (MRI) identified a large sellar enhancing lesion with cystic degeneration measuring 2.9 x 1.7 x 2.3 mm. (Figure 1).

Hormone profiles including cortisol, plasma adrenocorticotropic hormone (ACTH, thyroid hormones TSH and T4 follicle stimulating

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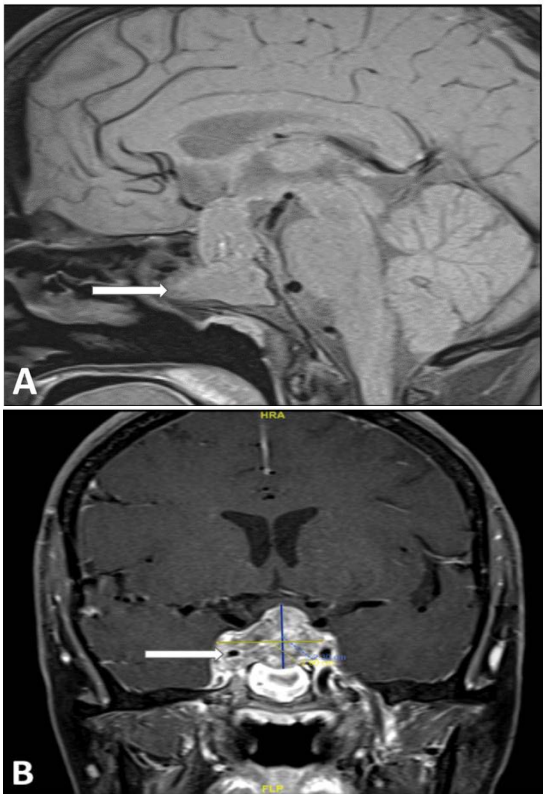

Figure 1: (a) T1-weighted MRI Sagittal, images show large intrasellar mass (arrow), (b)T1-weighted MRI Coronal, images show large intrasellar mass encasing right internal carotid artery (arrow).

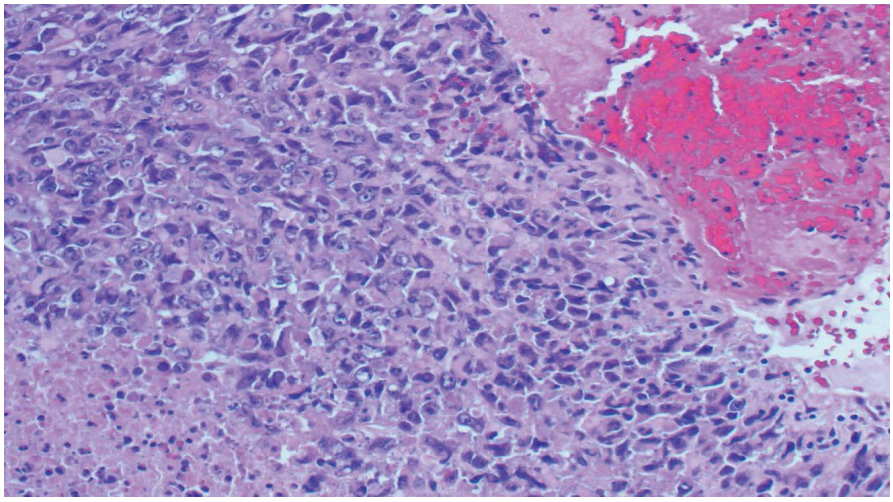

Figure 2: Tumor is composed of sheets of undifferentiated cells with large area of necrosis and hemorrhage (H&E stain, ×200 magnifications).

hormone (FSH), luteinizing hormone (LH) and Growth hormone, prolactin was normal.

Classical trans-nasal endoscopic approach surgery was started by ENT surgeons, after exposing the base of sella the neurosurgeon took over, dissection of the tumor from the right side, posterior and anterior was done. The tumor was rubbery and fibrotic which was unusual to pituitary adenoma. Shifting to left side of the cavernous, the pituitary was lying over the medial wall of the cavernous sinus.

The histopathological study revealed a high-grade densely cellular neoplasm (Figure 2). Tumor cells were large-sized, polygonal in shape and arranged in sheets. The cytoplasm was eosinophilic with focal eccentric eosinophilic globular inclusions. The nuclei were oval-shaped and pleomorphic with prominent nucleoli (Figure 3). First differential diagnosis was pituitary adenoma as it is the most common tumor in the sellar region. Other differential diagnosis would include glioma and atypical teratoid rhoboid tumors. Negative staining for pituitary

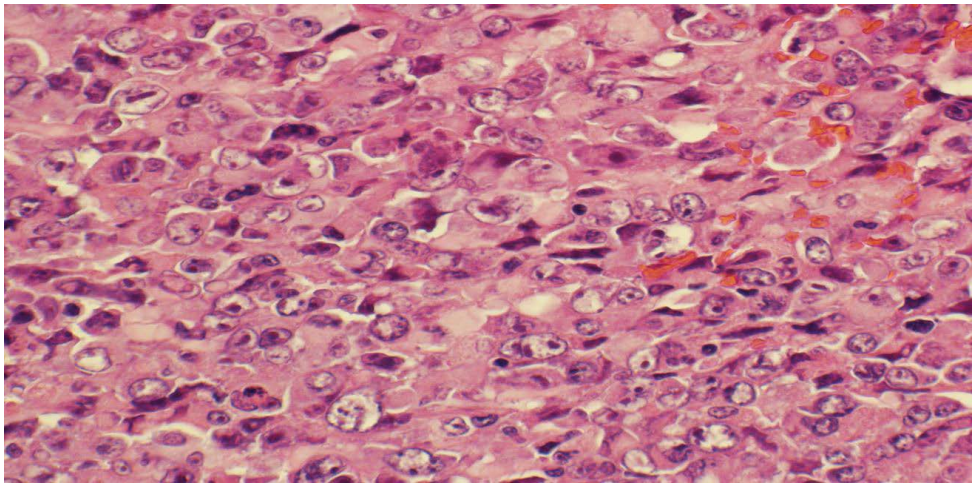

Figure 3: Tumor is composed of Medium sized to large cells with prominent nucleoli and some with peripherally placed nuclei. (H&E stain, ×400 magnifications).

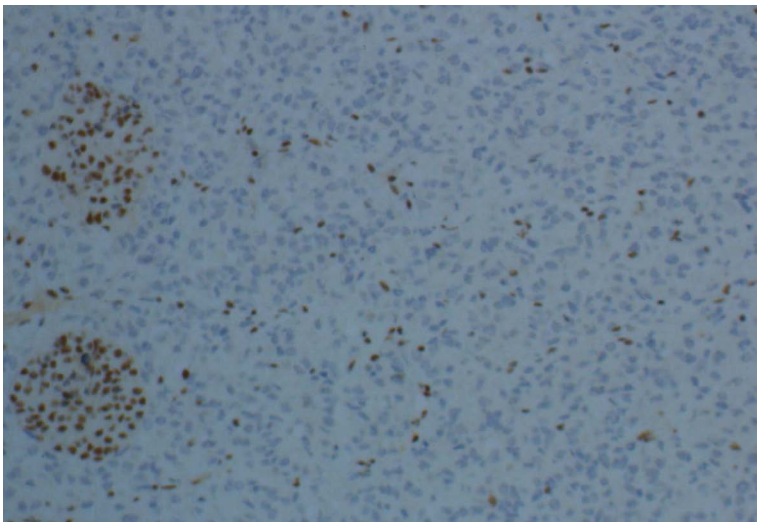

Figure 4: INI-1 (BAF-47) immunostaining shows loss of nuclear staining in the tumor nuclei and positive in the endothelial cells and residual intrapped pituitary tissue. (× 200 magnification).

panel, chromogranin and synaptophysine excluded pituitary adenoma and other neuroendocrine tumors as well. Glial fibrillary acidic protein (GFAP) was negative excluding glial tumor. Immunohistochemical stain for INI-1 (BAF- 47) was negative in the tumor nuclei (Figure 4). The morphologic characteristics and immunoprofile were diagnostic of AT/RT.

Post-operative she developed diabetes insipidus (DI) which was treated by desmopressin. She was for follow up with endocrinologist, she had complete ptosis. She was referred to oncology team, started on chemo-radiation therapy. One month later the patient died due to respiratory failure secondary to acute respiratory distress syndrome in shock state and acute kidney injury.

Discussion

Atypical teratoid rhabdoid tumors (AT/RTs) are aggressive pediatric malignant brain tumors that accounts for approximately 1-2% of pediatric cancers of the CNS [5]. The majority of these tumors (approximately 60%) occurs in the posterior cranial fossa (particularly the cerebellum) [6]. Only 45 cases of adults with AT/RT have been reported in the literature to date [5]. Ostrom et al. reported that the most common location in adults is the cerebral hemisphere, followed by the sellar region [7]. In contrast, Vivien et al. found that the most common location was the sellar region (46%), followed by cerebral hemisphere (32%) [8]. Sellar AT/RTs were reported to be almost exclusively in females and not in the pediatric population [9,10]. Recently, it has been reported in the sellar region, with only 14 cases reported in the literature to date including our case (Table 1) [4,6,11-19]. All fourteen cases are female of age range between (20- 61) years old, and most of the cases presented with visual disturbance and oculo- palsies.

Radiological findings of sellar AT/RT are non-specific but are remarkably similar to pituitary adenoma; the lesions are isointense on T1-weighted imaging and enhance following gadolinium administration [12]. Histologically, AT/RT are composed of diffuse proliferation of atypical large cells showing eccentrically located nuclei and abundant eosinophilic cytoplasm (rhabdoid features) with

Table 1: Summary of previously described of sellar AT/RTs and the present case

	Year	Age/Se x	Presentation	Treatment	Outcome	Reference
1	2000	32/F	Visual disturbance	Resection, Radiation, and Chemotherapy	28 months (dead)	[12]
2	2005	20/F	Visual disturbance	Resection, Radiation, and Chemotherapy	28 months (survived)	[13]
3	2005	31/F	Not mentioned	Resection, and Radiation.	9 months (dead)	[13]
4	2008	56/F	Diplopia	Resection, and Radiation.	23 months (dead)	[14]
5	2010	46/F	Headache	Not mentioned	Not mentioned	[15]
6	2011	61/F	Diplopia	Resection	3 months (dead)	[16]
7	2011	57/F	Diplopia	Resection, Radiation, and Chemotherapy	6 months (survived)	[16]
8	2013	60/F	Headache and diplopia	Resection, radiation, and chemotherapy	30 months (dead)	[17]
9	2013	43/ F	Headache and diplopia	Resection and radiation	2 weeks (survived)	[18]
10	2014	44 /F	Visual disturbance	Resection, radiation, and chemotherapy	17 months (dead)	[19]
11	2014	42/F	Visual disturbance	Resection, radiation, and chemotherapy	27 months (survived)	[5]
12	2015	36 /F	Headache and blurred vision	Resection, radiation, and chemotherapy	29 months (dead)	[20]
13	2017	35/ F	Headache, diplopia, and amenorrhea	Resection, radiation, and chemotherapy	37 months (survived)	[7]
14	Present case	40/F	Headache, right oculomotor palsy with decrease vision	Resection, radiation, chemotherapy	2 months	

prominent nucleoli [4]. SMARCB1/INI1 is one of the core subunit proteins of the ATP-dependent SWI/SNF chromatin remodeling complex, and is identified as a potent and bona fide tumor suppressor [20]. Interactions have been demonstrated between SMARCB1/INI1 and key proteins in various pathways related to tumor proliferation and progression: the p16-RB pathway, WNT signaling pathway, sonic hedgehog signaling pathway and Polycomb pathway [20]. The molecular changes in AT\RTs include a mutation in one allele with a second allele loss due to monosomy 22, deletion of 22q11.2, or an acquired copy number neutral loss heterozygosity [21, 22]. Sellar AT/RTs also have a higher prevalence of biallelic INI1 alterations compared to AT/RTs in other locations [10]. Loss of expression of INI1 as detected by immunohistochemical staining correlates with deletion and mutations of the INI1 gene [23]. The differential diagnosis of sellar mass includes pituitary adenomas (85%), followed by craniopharyngioma (3%), rathke cleft cyst (2%), meningioma (1%) [24]. Pituitary adenoma shows uniform nuclear morphology with moderately abundant cytoplasm, and the neoplastic cells are highlighted by synaptophysin and chromogranin. Craniopharyngioma have a distinct morphology that the neoplastic cells are forming nodules or trabeculae of squamous epithelium, with peripheral nuclear palisading that surrounds looser plumper cells called "stellate reticulum". In addition, nodules of anucleated squamous ("ghost" cells) and "wet" keratin are usually present. Rathke cleft cyst is a cyst lined by columnar ciliated epithelium with goblet cells. Meningioma usually have syncytial cells that have round uniform nuclei with intranuclear pseudo inclusions.

The prognosis of AT/RT is poor both in adults and in the pediatric population, Vivien et al, report the average survival rate of adult AT/RT is 20 months [8]. This is comparable to the reported median survival of 13.5 to 16.8 months in the pediatric population [10,25]. Long-term survival is possible in adult AT/RT cases after a combined approach including surgery, adjuvant radiotherapy, and chemotherapy [26, 27] (Table 1).

Conclusion

AT/RT is not limited to pediatric age group, it should be considered in the differential diagnosis of malignant sellar lesion in adult patients.

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This study is not funded.

Conflicts of Interest

The authors declare that they have no conflicts of interest.

Ethical Approval

This article does not contain any studies with human participants performed by any of the authors.

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