Intraventricular Superatentorial AT/RT in pediatrics : a case report and literature review

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Case Report

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Abstract

Introduction: Atypical teratoid/rhabdoid tumors (AT/RTs) are rare highly malignant and rapidly growing tumors usually seen in the posterior fossa in children and less commonly supratentorial compartment. Intraventricular location is considered even more scarce. The authors report a case of ATRT originating from the lateral ventricle in a child and did a literature review

Methodology: To investigate for previous reports of an atypical tertatoid and rhaboid tumors located in the lateral ventricle in children, we performed a literature search that yielded 19 results. These articles were screened for cases of primary supratentorial intraventriuclar AT/RTs.

Result: The literature search led to the identification of 11 articles including 11 patients. Herein, the authors present the 12th case of AT/RT in a 5 yr old who presented with vomiting and generalized tonic clonic seizures. Computed tomography scan and magnetic resonance imaging showed heterogeneous huge mass within the left lateral ventricle with central area of hemorrhage and necrosis . The patient underwent Emergency left frontal craniotomy with complete removal of the tumor. Histological examination confirmed the diagnosis of AT/RT

Conclusion: The results of a literature search suggest that a ATRT arising in the lateral ventricle is quit rare. These cases emphasize on the necessity of considering AT/RT in the list of differential diagnosis of supratentorial intraventricular tumors in pediatrics. The prognosis of this particular subtype is still poor.

Introduction

AT/RTs (atypical teratoid/rhabdoid tumors) are highly aggressive, quickly growing tumors with extensive vascularity that are frequently linked with cerebrospinal fluid (CSF) seeding and a poor prognosis. [2] It is classified as a grade IV neoplasm by the World Health Organization. [6] Due to histological and radiological similarities, AT/RTs have been misdiagnosed as primitive neuroectodermal tumours/medulloblastomas (PNETs/MBs) in the past.[5] Supratentorial ATRT's are uncommon in children, and intraventricular location is considered extremely rare.[1] There are currently no guidelines for the best management of AT/RT. Herein, we present a case of lateral ventricle AT/RT in a child. Furthermore, take a look at the literature. The goal is to draw attention to this tumor unusual location and radiological findings.

Literature review :

A search through MEDLINE using the MeSH keywords "AT/RT" OR "Atypical teratoid/rhabdoid " AND "intraventricular." yielded 19 results. Articles referencing tumors located infratentorially, or intraparynchymal were excluded. Articles potentially meeting criteria for inclusion on the basis of title and abstract were reviewed and reports of pathologically confirmed AT/RT arising within the ventricles supratentorially were included. A Flow diagram demonstrating the systematic analysis process [Figure 1]. Similar to our case, the majority of these reports (5 cases) originated within the lateral ventricle.[1, 5, 8,

11] ,while two cases were described involving the temporal horn [10, 4]. Three cases were reported in the septum palladium [6, 7, 11] and only one case in the Velum interposium. [Donovan] Results are summarized in [Table 1]. Our search demonstrated only one case of superatentorial intraventriuclar AT/RT in adults. [12]

Case Report

We report the case of a 5-year-old boy who was seen in the clinic complaining of headache. However, he presented to Emergency department with episode of vomiting and generalized seizure. His Glasgow comma score was fluctuating between 8 to12 and his Pupils were unequal. A head Computed Tomography (CT) was obtained and revealed a large intra-ventricular tumor occupying the left lateral ventricles with intra-tumoral bleeding causing obstructive hydrocephalus. His Magnetic Reasoning Images (MRI) done before presentation showed a huge lobulated lesion with heterogeneous signal intensity, necrotic area, and contrast enhancement, involving the inner wall of the left lateral ventricle down to upper anterior third ventricle, The radiological appearance of the lesion was suggestive of Intraventricular Primitive Neuroectodermal Tumor (PNET), or Anaplastic Astrocytoma [Figure 2].

An emergency left forntoparital craniotomy and transcortical approach was performed to obtain diagnostic tissue and tumor debulking. The tumor was bloody, necrotic and fragile, tumor resection was done and bilateral External ventricular drains were inserted. postoperatively, the patient developed expressive aphasia and right sided facial weakness and limb hemiparesis.

Histopathology showed pleomorphic rhabdoid cells with abundant eosinophilic cytoplasm, with filamentous cytoplasmic inclusions and vacuoles; as well as eccentric round nuclei and prominent nucleolus. IHC demonstrated tumor cells to be positive for epithelial membrane antigen (EMA), with focal positivity for smooth muscle antigen (SMA), and Synaptophysin. It also revealed a lack of nuclear integrase interactor 1 (INI-1) in tumor cells, with a Ki-67 of 25–30% thus confirming the diagnosis of AT/RT WHO grade IV tumor. [Figure 3]

Postoperative imaging showed a small non-enhancing residual, diffuse cranial nerves and leptomeningeal nodular enhancement suggestive of disseminated disease.The case was discussed in the multidisciplinary pediatric Neuro-Onoclogy tumor board and he later received first cycle of chemotherapy but Unfortunately the patient passed away 3 months after the primary surgery.

Discussion

ATRT’s account for approximately 1.3 percent of all pediatric CNS tumors and up to 20% of all embryonal tumors. [13] the largest and most recent series in the literature reported 62% of tumors occurring supratentorialy while only 38% in infratentorial location. [2] Supratentroial ATRT’s typically arise in the parenchyma of the cerebral hemisphere; those originating in the suprasellar and pineal areas may directly involve the ventricles, but primary intraventricular atypical teratoid/rhabdoid tumors are extremely rare. [2,

10] We provide an instance of pathologically verified AT/RT in the lateral ventricles in this article. To the best of our knowledge, and based on our literature search, an AT/RT occurring supratentorially inside the ventricles has only been described in 11 cases in the literature.

Molecular studies have revealed a pathognomonic alteration of a tumor suppressor gene in AR/RT : The INI1/hSNF5 gene on chromosome 22 also known as SMARCB1. [4] this mutation is absent in medulloblastoma and PNET tumors but may be present in other tumors such as choroid plexus carcinoma.[4, 6] It has become the gold standard approach for distinguishing this entity from the previously stated morphologically comparable malignancies.[6]

Although the imaging findings of AT/RT are often ambiguous, presence of hemorrhage and meningeal dissemination are highly suggestive. [1] In our case, heterogeneous enhancement of the tumor with intra-tumoral bleeding and necrosis as well as CSF dissemination was evident. Because of the difficulty in accessing and managing the tumor vasculature within the tumor's center, the location reported provides a unique challenge. In our case, a transcortical technique was used, which resulted in significant bleeding before the arteries could be isolated. There are currently no particular guidelines established for the management of AT/RTs. Multimodal therapy including surgical resection, radiation, and chemotherapy are all part of the treatment strategy. [3] One of the most important criteria in predicting prognosis appears to be the degree of surgical resection. [2, 9] The patients' age and incapacity to get full dose craniospinal irradiation are also key factors in their prognosis. Nonetheless rapid disease recurrence and progression are common even in those who receive full dosage craniospinal irradiation.[2, 6] Overall, the prognosis is poor, with a median survival time of only six months.[4, 5]

Conclusion

Supratentorial AT/RT is uncommon in children and location in the lateral ventricle is seldom described in the literature. These cases are intriguing because of their odd location and nonspecific imaging findings that resemble other childhood tumors. Despite multimodal treatment options, these tumors remains highly malignant, with a high rate of morbidity and mortality, elucidating the dismal prognosis. Given its rarity and poor outcome, this enigmatic tumor should be kept in mind when considering the differential diagnosis of malignant Intraventriuclar brain tumors in pediatrics.

Declarations

All authors certify that they have no affiliations with or involvement in any organization or entity with any financial interest or non-financial interest in the subject matter or materials discussed in this manuscript and there’s no Competing interests.

Ethics approval and consent to participate

This case report was approved by the The Office of Research Affairs in king faisal specialist hospital and research center , assigned with RAC number: 2225092.

Consent for publication : 'Not applicable' our case report does not disclose any patient information or identification, the parents verbally consented to publish the case with no disclosure of patient identity or information, thus a written consent was not obtained

Availability of data and materials : data published in the paper are presented and further details can be provided upon request

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Authors contribution :

All authors contributed to the study conception and design.

Material preparation were performed by Thana Namer ,Maryam Alotaibi and Othman Alhammad.

Data collection and analysis was performed by Thana Namer

The first draft of the manuscript was written by Thana Namer and all authors commented on previous versions of the manuscript.

All authors read and approved the final manuscript.

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Tables

Table.1 : Reported case of Supratentorial Intraventricular ATRT

Author	Age	Gender	location	Drop mets	Treatement	Outcome
Ahmed et. Al 2005 [11]	4yrs	F	Left lateral ventricle	N/A	GTR + CR	Death in 10mo.
Parwani et. Al 2005	5 yrs	M	Left lateral ventricle	N/A	GTR	N/A
Donovan et. Al 2006	11 wks	F	Velum interpositum	Negative	1- STR 2- GTR + C	No recurrence for 4 years.
Meyers et.al 2006	4yrs	F	Septum pellucidum	N/A	GTR + CR	N/A
Lee et.al 2009	1.4 yrs	M	Lateral ventricle	Negative	GTR	N/A
Li et.al 2012	3 yrs	M	Septum pellucidum	N/A	GTR + CSR	Died 2 years after surgery.
Feng et.al 2013 [11]	3 yrs	F	Septum pellucidum	N/A	GTR + CSR	Death.
Darmoul et.al 2015	2 mo.	M	Left lateral ventricle	Negative	GTR + C	No recurrence for 3 years.
Singh et.al 2016	4 yrs	M	Right temporal horn	N/A	GTR + CSR+ C	Developed multiple poster fossa mets at 2mo. , Died 3mo. after surgery
Sharma et.al 2020	4yrs	M	Left lateral ventricle	N/A	GTR + CSR	N/A
Lakhder et.al 2020	4 yrs	M	Entire temporal form and the body of the left lateral ventricle	Negative	GTR then GKS after recurrence + CR	Recurrence twice , died after 1 year.
Current case	5 yrs	M	Left lateral ventricle	Diffuse leptomeningeal enhancement	GTR + CSR	N/A

GTR (gross tumor removal), C (chemotherapy), R (radiation therapy), CSR(craniospinal radiation) NA (not available).

Figures

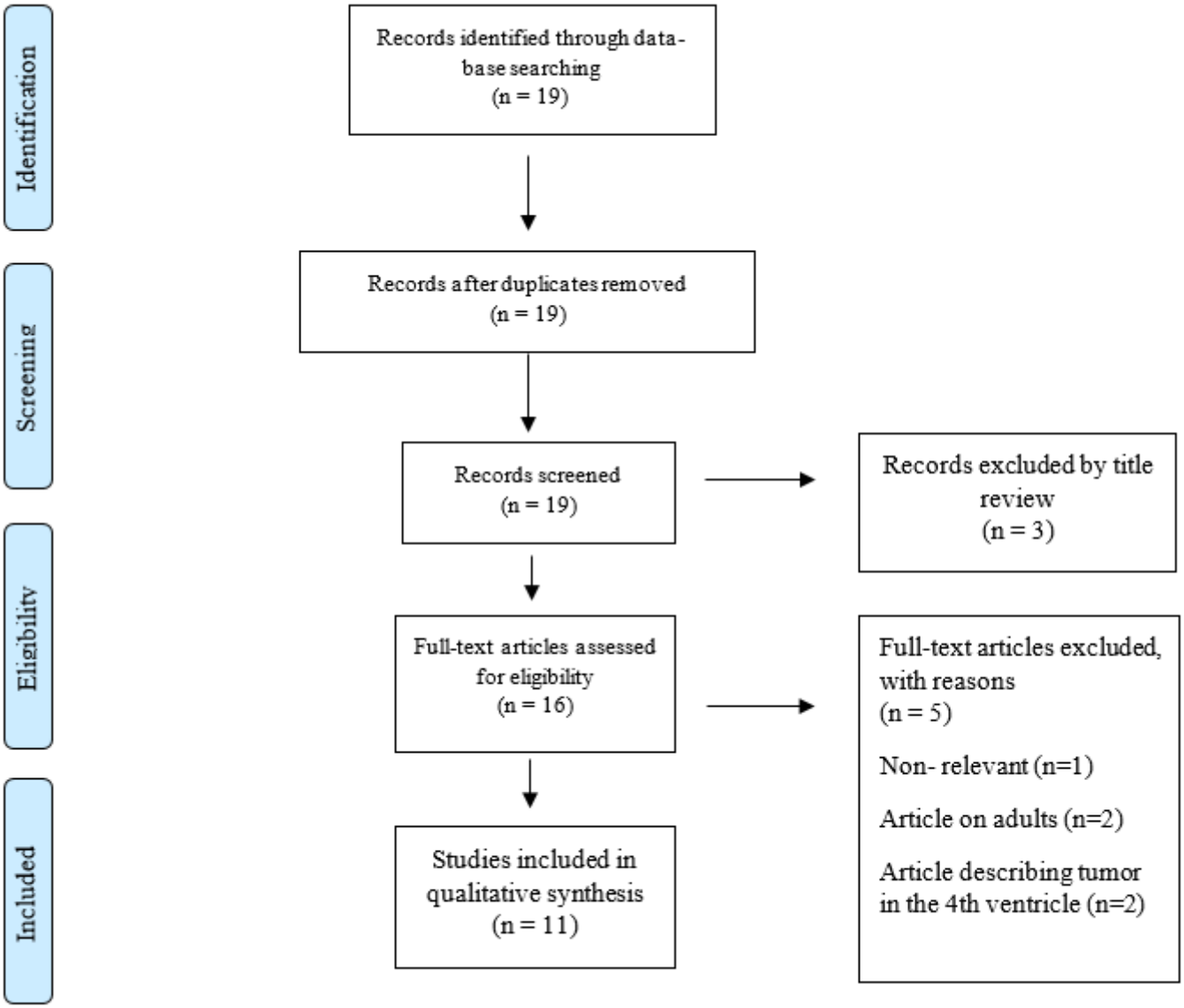

Figure 1

Flow diagram demonstrating the systematic analysis process

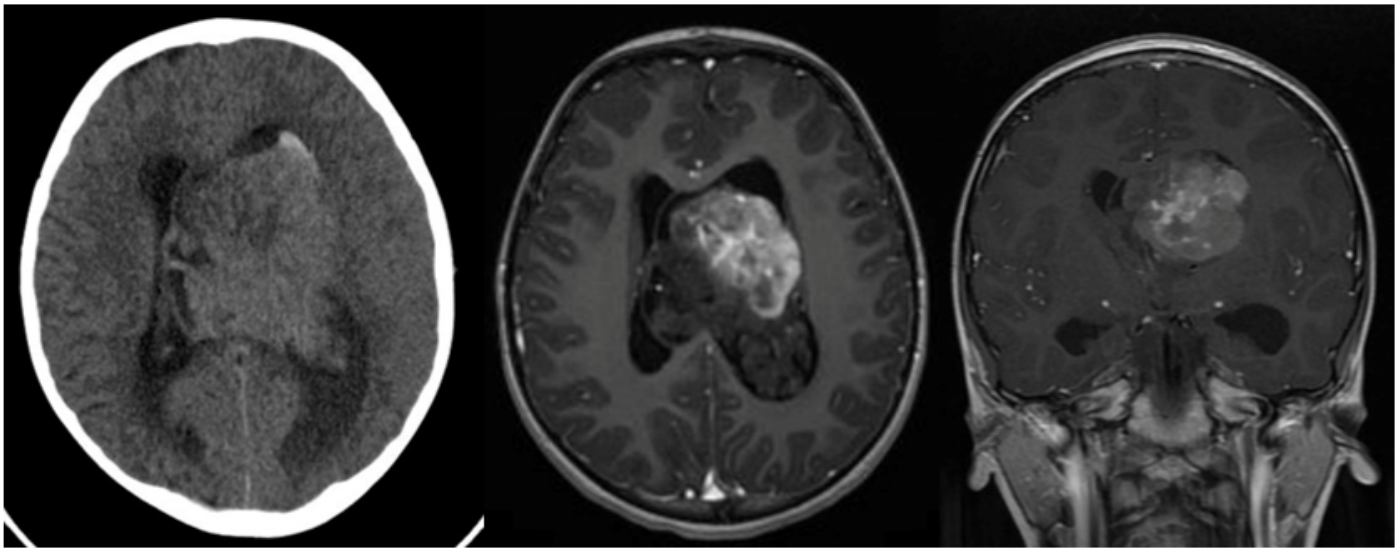

Figure 2

Preoperative CT and MRI. (a) Large intra-ventricular tumor occupying the left lateral ventricles causing obstructive hydrocephalus. (b,c) Axial and coronal T1-weighted post-gadolinium image showing a huge lobulated lesion (measuring approximately 48 × mm 47 mm × 57 mm), with heterogeneous signal intensity, necrotic area, and contrast enhancement, involving the inner wall of the left lateral ventricle down to upper anterior third ventricle, obstructing the foramina of Monro and causing hydrocephalus.

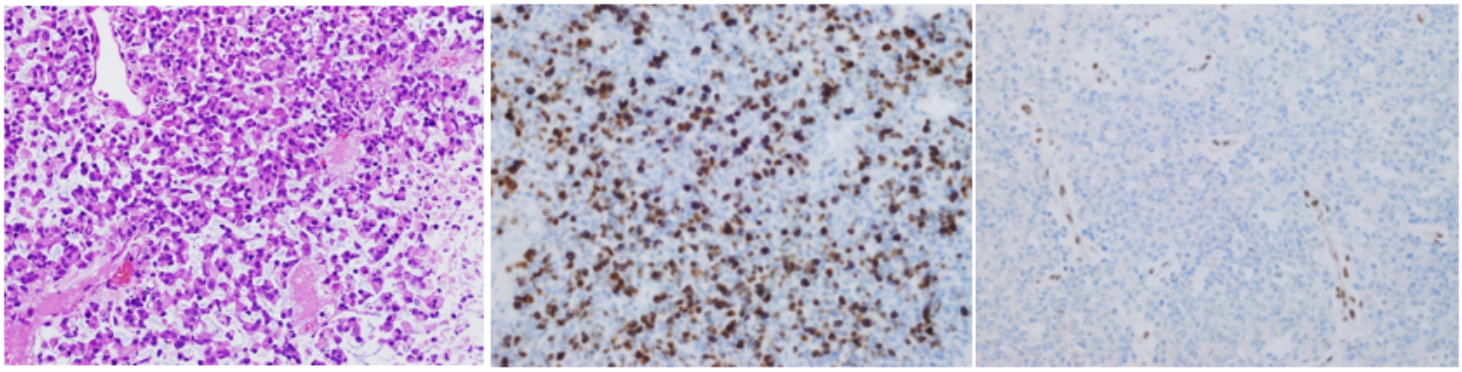

Figure 3

(A) Hematoxylin and eosin (H&E) stain showing rhabdoid cells with vesicular chromatin, prominent nucleoli, eosinophilic globular cytoplasmic inclusions with abundant eosinophilic cytoplasm. (b) Ki-67 staining showed Ki-67 index was 25-30%. (c) Loss of expression of SMARCB1 (INI1) in nuclei of tumour cells, was found with retained expression in Intratumoural blood vessels.