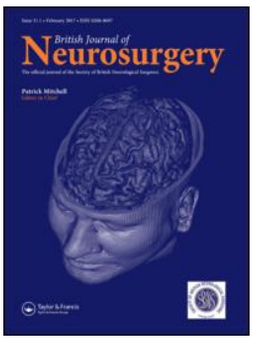

British Journal of Neurosurgery

ISSN: (Print) (Online) Journal homepage: https://www.tandfonline.com/loi/ibjn20

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To cite this article: Xuanyu Tan, Mengliang Xiao, Wei Jiang, Cao Yue, Limei Qu & Gang Zhao (2021): Paediatric atypical teratoid/rhabdoid tumour of the pineal region mimicking a meningioma: a case report and literature review, British Journal of Neurosurgery, DOI: 10.1080/02688697.2021.1895971

To link to this article: https://doi.org/10.1080/02688697.2021.1895971

Published online: 16 Mar 2021.

BRITISH JOURNAL OF NEUROSURGERY
https://doi.org/10.1080/02688697.2021.1895971

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SHORT REPORT

Paediatric atypical teratoid/rhabdoid tumour of the pineal region mimicking a meningioma: a case report and literature review

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ABSTRACT

Purpose: Atypical teratoid/rhabdoid tumour (AT/RT) is a highly malignant central nervous system tumour of early childhood. According to the latest WHO classification, the diagnosis of AT/RTs needs to be confirmed by the absence of SMARCB1 (INI1) or SMARCA4 (BRG1) protein expression. AT/RT in the pineal region is infrequent and most have not been proven genetically. Here, we report a case of AT/RT in the pineal region, preoperatively misdiagnosed as a meningioma. Immunohistochemistry revealed the absence of INI1 protein expression.

Method: A 29-month-old boy was admitted to the hospital after 14 days of emotional apathy and a 2-day vomiting history. AT/RT was not considered during the initial diagnosis because this tumour is rare in this region and is often accompanied by cystic degeneration and necrosis on imaging. Subsequently, the patient underwent surgery and the tumour was completely excised.

Result: The pathological diagnosis was AT/RT. After discharge, the patient continued chemotherapy in other hospitals but died five months after surgery because of disease progression.

Conclusion: To our knowledge, this is the fifth case of paediatric pineal AT/RT confirmed genetically. Although in children AT/RT in the pineal gland is rare, a differential diagnosis of AT/RT should be considered when new pineal masses appear in children. For this highly malignant disease with poor prognosis, it is very important to detect and recognize the disease as soon as possible, and to adopt surgery plus multiple treatment management.

ARTICLE HISTORY
Received 15 September 2020
Revised 17 February 2021
Accepted 19 February 2021

KEYWORDS
Paediatric; pineal region; atypical teratoid/rhabdoid tumour; neurosurgery

Introduction

Pineal region tumours account for less than 10% of children's brain tumour cases.[1] They are a group of tumours with high histopathological heterogeneity, the most common being germ cell tumours (GCTs), followed by pineal parenchymal tumours (PPTs).[2] Atypical teratoid/rhabdoid tumours (AT/RTs) are highly malignant and are classified as a grade IV tumour by the WHO. AT/RTs constitute approximately 2% of all paediatric central nervous system tumours.[3] This tumour often occurs in the posterior fossa with only rare studies reporting cases of AT/RTs in the pineal region. Only a few of these cases have been genetically confirmed. AT/RTs are characterized by an inactivation or mutation of the *INI1* gene on chromosome 22q11.2. According to the latest WHO classification of central nervous system tumours, the diagnosis of AT/RTs must be confirmed by the above gene mutation (or the rarer *BRG1* mutation). Without this gene mutation, and based on only histologically consistent AT/RTs findings, these tumours can only be descriptively diagnosed as central nervous system embryonal tumours with rhabdoid features.[4]

Here we report a case of AT/RTs in the pineal region of a boy. Immunohistochemistry revealed the loss of INI1 protein expression in the nucleolus. To the best of our knowledge, this is the fifth case of AT/RTs in the pineal region confirmed genetically. We collated the imaging and clinical findings, the treatments initiated, and conducted a brief literature review.

Case presentation

Patient history and examination

A 29-month-old boy was admitted to the hospital after 14 days of emotional apathy and 2-day vomiting history. The patient had a history of a fall with a head injury a week prior. The patient's parents observed that the child experienced loss of appetite and weight. Physical examination revealed no sensory or motor disorders. The pupillary reflex and eye movement were normal. Serum tumour markers (including the alpha-fetoprotein, carcinoembryonic antigen, human chorionic gonadotropin, etc.) were analysed preoperatively, with only neuron specific enolase being slightly raised.

Neuroimaging findings

MRI showed a solid tumour in the pineal region measuring approximately $5.2 \times 4.5 \times 4.0$ cm. The tumour appeared with a low signal intensity on T1-weighted MRI (Figure 1(A)) and hyperintense on T2-weighted MRI (Figure 1(B)) when compared with the white matter. There was hydrocephaly in the lateral and third ventricles caused by tumour compression of the aqueduct. There was oedema of the white matter around the occipital angle of the lateral ventricle (Figure 1(B)). The tumour showed heterogeneous enhancement (Figure 1(C–E)). The third ventricle and

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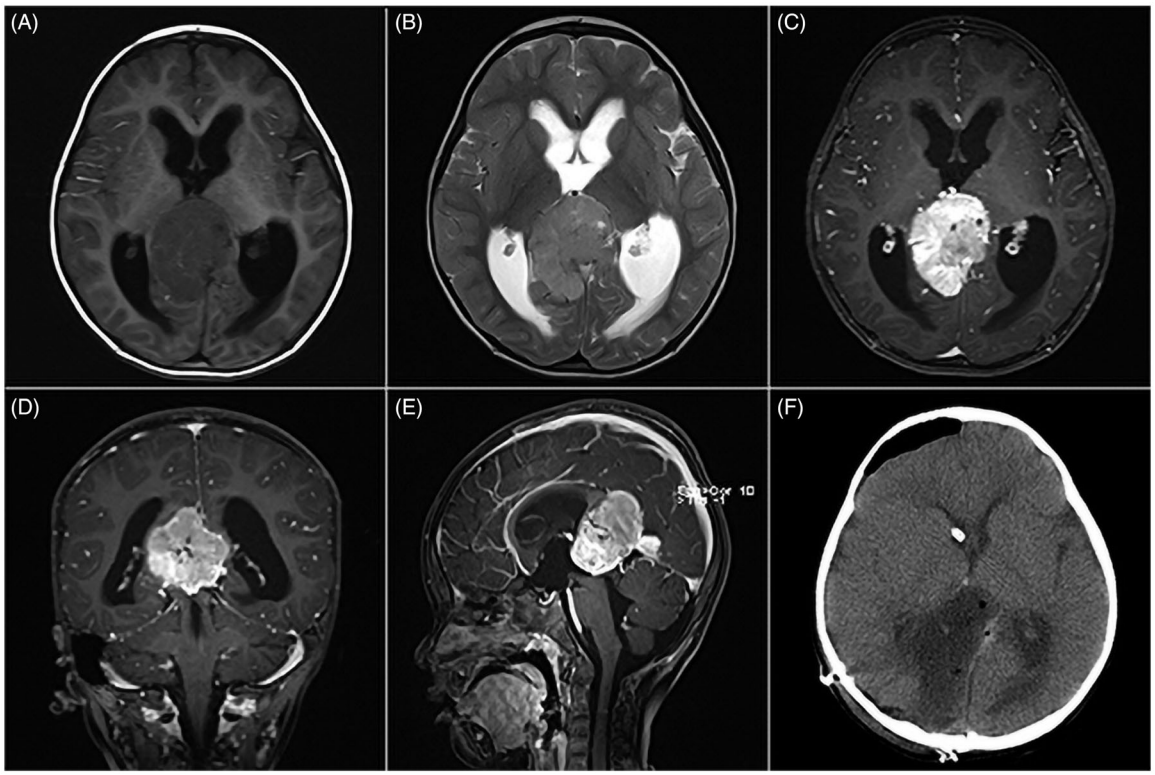

Figure 1. Preoperative and postoperative imaging. A large lobulated solid mass located in the pineal region, compressing the third ventricle and corpus callosum. (A,B) The tumour appears with a low signal intensity on T1-weighted MRI and hyperintense on T2-weighted MRI when compared with the white matter. Lateral and third ventricles are dilated. (C–E) Axial imaging, coronal imaging, sagittal images with administration of the contrast medium, the tumour showing obvious heterogeneous enhancement. (F) Five hours after the first surgery, head axial computed tomography showing complete removal of all the tumours.

corpus callosum were deformed by tumour compression (Figure 1(E)). According to the imaging findings and laboratory results, it was initially diagnosed as a meningioma.

Surgery and postoperative course

The patient was placed on the right side, and the surgery used the Poppen approach. The bone window was aligned with the midline and the transverse sinus. The focus was on fully exposing the sagittal and transverse sinus. During surgery, the tumour surface was relatively smooth, and the boundary was clear. The tumour compressed the third ventricle, corpus callosum, and midbrain. After removing the tumour in the capsule, we carefully separated the periphery of the tumour to protect the brain tissue and blood vessels. The tumour was greyish white, solid, and slightly tough. To prevent postoperative hydrocephalus, during the surgery, an external ventricular drain was placed. Finally, the tumour was completely excised under the microscope. The postoperative state of the patient was relatively stable, and no related complications occurred. Postoperative computed tomography showed complete tumour removal (Figure 1(F)). After discharge, the patient continued chemotherapy in other hospitals but died five months after surgery because of disease progression.

Pathological findings

Most tumour cells were composed of rhabdomyoid cells (Figure 2(A)). These cells have clear cell boundaries, deviated nuclei, obvious nucleoli, abundant eosinophilic cytoplasm, and extensive vacuoles in the cytoplasm. Immunohistochemical examination revealed local positivity for epithelial membrane antigen (Figure 2(B), ×200), cytokeratin (Figure 2(C), ×200), glial fibrillary acidic protein (Figure 2(D), ×200), and smooth muscle actin (Figure 2(E), ×200). INI1 expression was negative (Figure 2(F), ×200); therefore, the final pathological diagnosis was AT/RT.

Discussion

Although AT/RT can occur in any part of the nervous system, approximately 50% of cases occur in the posterior fossa.[5–7] So far, only four cases of pineal AT/RT in children confirmed by genetics have been reported in English literature (Table 1). Male patients are slightly more affected, with the ratio of male to female approximately 1.5:1.[10] Notably, the four cases previously reported in the literature are all female patients, while the present case is of a male patient. Because of its malignancy, AT/RT can spread and travel via cerebrospinal fluid in the early stages, with 30–40% of patients presenting with metastases at the time of detection.[11]

The clinical manifestations of AT/RT are related to tumour location and are, therefore, not specific. In our patient, similar clinical manifestations to other pineal tumours, tumour obstruction or compression of the midbrain aqueduct led to hydrocephalus and an increased intracranial pressure with patients often complaining of headache, nausea, vomiting and dizziness.[5,12]

The imaging features were nonspecific, but there are similar findings in some reported cases.[13,15] AT/RT often shows an inhomogeneous enhancement after contrast agent injection. Most tumours are accompanied by cystic, necrotic, and haemorrhagic areas.[12,15,16]Approximately, 40% of patients have an unusual MRI finding showing an annular band around the cystic or necrotic area in the middle.[16] However, our case is unusual in that these

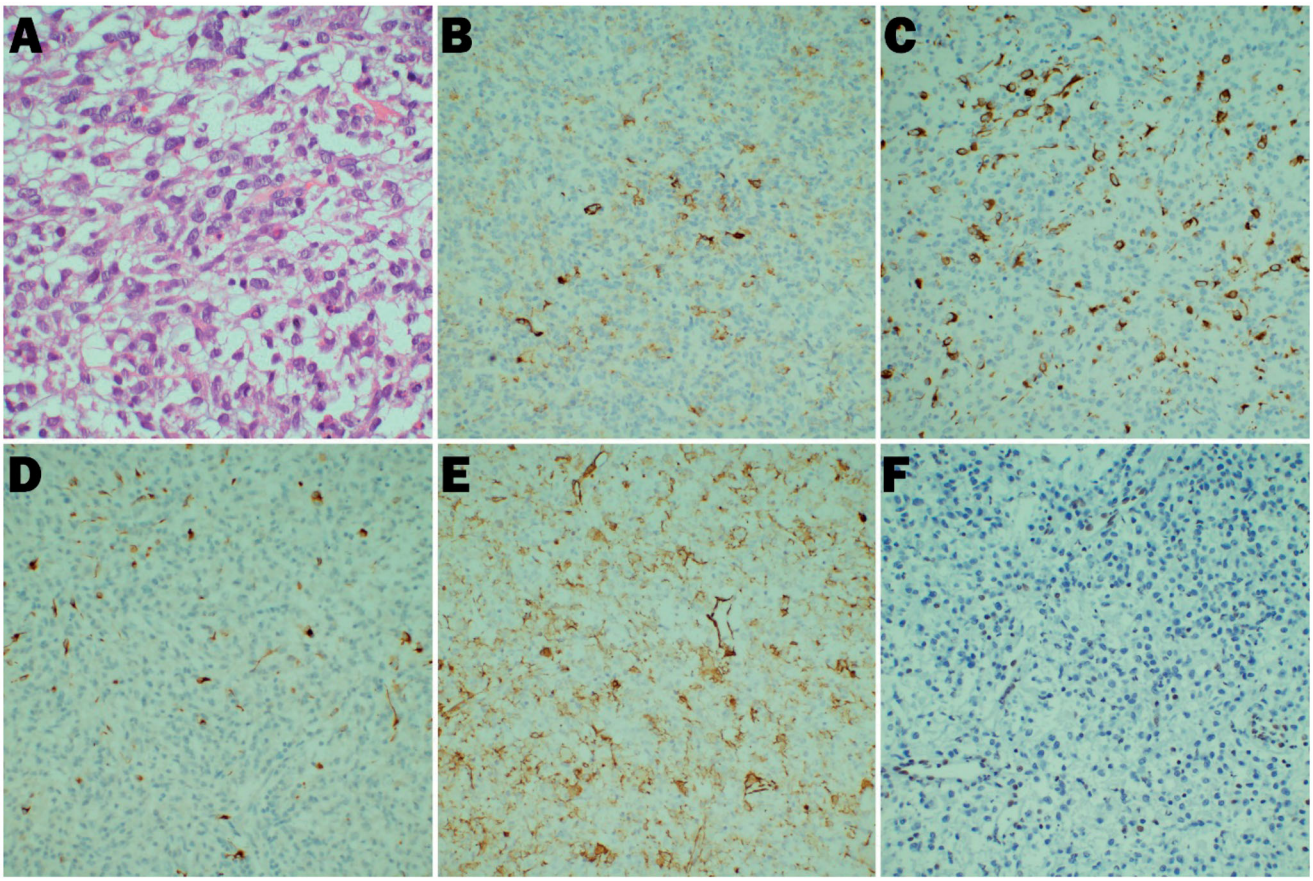

Figure 2. Histopathological examination of the resected specimen. (A, magnification ×400) Most tumour tissues are composed of rhabdomyoid cells. (B–E, ×200) Immunohistochemical examination showing local positivity for epithelial membrane antigen, cytokeratin, glial fibrillary acidic protein, and smooth muscle actin. (F, ×200) The expression of INI1 is negative.

Table 1. Summary of paediatric atypical teratoid / rhabdoid tumours in the pineal region.

Author	Sex/age (month)	Symptom	INI1	Immunostaining	Treatment	Outcome
Wang *et al.*[8]	F/23	1-month history of gait unsteadiness and 10-day history of headache, nausea, and vomiting.	–	Vim EMA CK CD99 CD56	GTR	Relapsed, 3 months after surgery
	F/26	2-week history of headache and vomiting	–	Vim EMA CD56 CD99 nestin	GTR	Died, 5 months after operation
Yang *et al.*[9]	F/17	Drowsiness, vomiting, coma	–	Vim EMA GFAP SMA	GTR	Died, 8 onths after operation
	F/40	Headache, vomiting	–	Vim EMA GFAP S-100 CK	STR	Died, 4 months after operation

GTR: gross total resection; STR: subtotal resection.

common imaging features were not observed. Therefore, it was initially diagnosed as a meningioma because of its close relationship with the dura mater and obvious enhancement. Based on the results of our reported case, even if atypical imaging is present, in young children, we should pay attention to the possibility of AT/RT.

Conclusion

Although AT/RT in the pineal gland is rare in children, a differential diagnosis of AT/RT should be considered when new pineal masses appear.

Ethical approval

Written informed consent for publication of their details was obtained from the patient's parents.

Acknowledgement

This study did not receive any funding.

Disclosure statement

No potential conflict of interest was reported by the author(s).

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