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Case Report

Pediatric Atypical Teratoid/Rhabdoid Tumor of the Cerebellum: A Case Report and Literature Review

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Abstract

Atypical teratoid/rhabdoid tumor (AT/RT) is a rare and highly aggressive embryonal tumor that predominantly affects infants and young children. This malignancy arises from primitive neuroectodermal cells and exhibits heterogeneous differentiation into various embryonic tissues. Due to its rarity and complexity, diagnosing and managing AT/RT present significant challenges. Recent studies have summarized the key features of cerebellar and supratentorial AT/RT cases; however, critical gaps remain in understanding their diffuse leptomeningeal variants and long-term functional outcomes. Here, we report a case of a two-year-old child diagnosed with cerebellar AT/RT, who presented with vomiting and gait instability. The patient underwent a gross total resection followed by adjuvant radiotherapy and chemotherapy. Despite achieving radiological remission, the patient survived for only eight months and experienced severe neurological deficits, including persistent ataxia and recurrent infections. This case highlights the disconnect between surgical success and long-term quality of life. It underscores the importance of integrating molecular diagnostics and palliative care to address the multifaceted burden of AT/RT.

Introduction

Atypical teratoid/rhabdoid tumor (AT/RT) is one of the most aggressive central nervous system (CNS) malignancies in pediatric patients, accounting for approximately 1–2% of all pediatric brain tumors.[1] It predominantly affects children under the age of three, with a slight male predominance.[2] AT/RT is characterized by rapid progression, resistance to conventional therapies, and poor prognosis, with a median survival of less than one year despite aggressive treatment.[3] The tumor typically arises in the posterior fossa, particularly in the cerebellum, but can also occur in supratentorial regions.[4] Histologically, AT/RT is composed of undifferentiated small round cells with rhabdoid features and often shows heterogeneous differentiation into mesenchymal, epithelial, and neural tissues.[5] Immunohistochemically, the tumor frequently expresses vimentin and other intermediate filaments, reflecting its neural crest origin.[5]

The molecular pathogenesis of AT/RT involves the inactivation of the SMARCB1 (INI1) or SMARCA4 (BRG1) genes, which are critical components of the SWI/SNF chromatin remodeling complex.[6] These genetic alterations lead to dysregulation of cell cycle control and differentiation, contributing to the tumor's aggressive behavior.[7] Despite advances in molecular diagnostics and multimodal therapies, including surgery, radiation, and chemotherapy, the prognosis for AT/RT remains dismal.[7] This case report aimed to provide a detailed account of the clinical presentation, diagnostic challenges, surgical management, and postoperative outcomes in a two-year-old girl with cerebellar AT/RT, contributing to the limited literature on this rare and devastating disease.

Case presentation

A two-year-old girl presented to Wuhan Union Hospital in November 2023 with a one-week history of frequent vomiting and gait instability. Brain magnetic resonance imaging (MRI) revealed a large mass in the fourth ventricle, accompanied by supratentorial hydrocephalus (Fig. 1). On physical examination, the patient was confused, with bilaterally equal and round pupils measuring 3 mm in diameter and reactive to light. Muscle strength in all four limbs was graded as 3. Following admission, comprehensive evaluations were completed, and no contraindications to surgery were identified. A posterior midline approach was employed to resect the cerebellar lesion. Intraoperatively, the tumor appeared slightly firm and fleshy, with moderate vascularity. Portions of the tumor were poorly demarcated from the inferior vermis of the cerebellum, while anteriorly it compressed the brainstem with clearly defined margins. Superiorly, it obstructed the aqueduct of Sylvius. After a three-hour procedure, gross total resection of the

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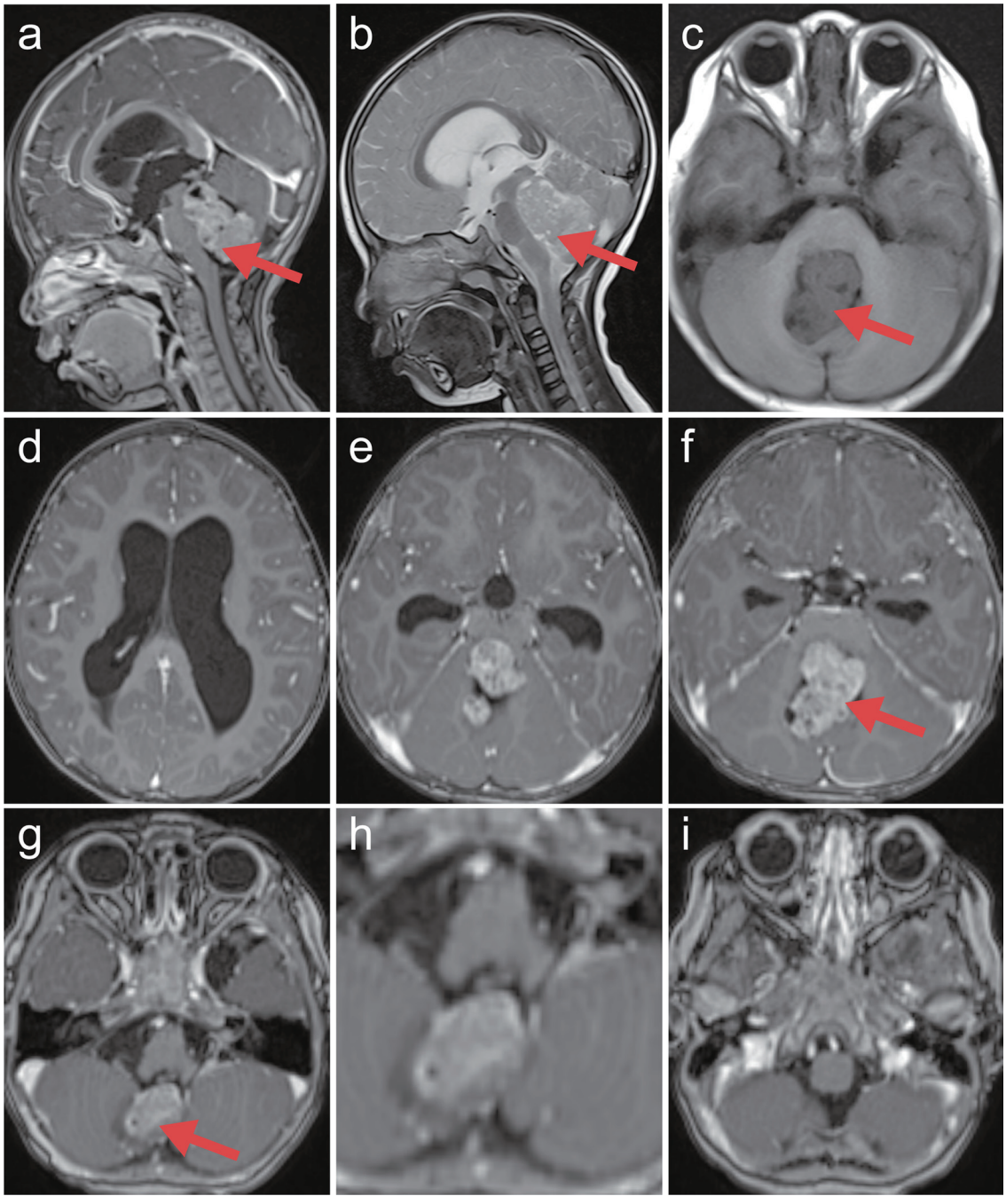

Fig. 1. Preoperative imaging findings of the atypical teratoid/rhabdoid tumor. (a) T1-enhanced presentation of tumors in the sagittal plane. (b) T2 manifestation of tumors in the sagittal plane. (c) T1 manifestation of tumors in the horizontal plane. (d–g, i) Imaging manifestations of tumors on continuous T1-enhanced scans. (h) Localized magnification indicates the unclear boundary between the tumor and the cerebellum. The red arrow indicates the tumor.

tumor was achieved.

Postoperative care and rehabilitation

Following surgical resection, the patient was closely monitored in the intensive care unit for 24 h to manage potential complications, such as intracranial pressure fluctuations and cerebrospinal fluid leakage. Postoperative imaging confirmed complete tumor removal (Fig. 2), and the patient was transferred to the pediatric ward for continued recovery. Over the subsequent two weeks, the patient underwent physical therapy to address gait instability and muscle weakness. Despite these efforts, the patient continued to experience significant neurological deficits, including persistent ataxia and impaired fine motor function. Psychological support was provided to the family to assist in coping with the emotional and physical challenges posed by the illness. The patient was discharged after three weeks with a plan for outpatient rehabilitation

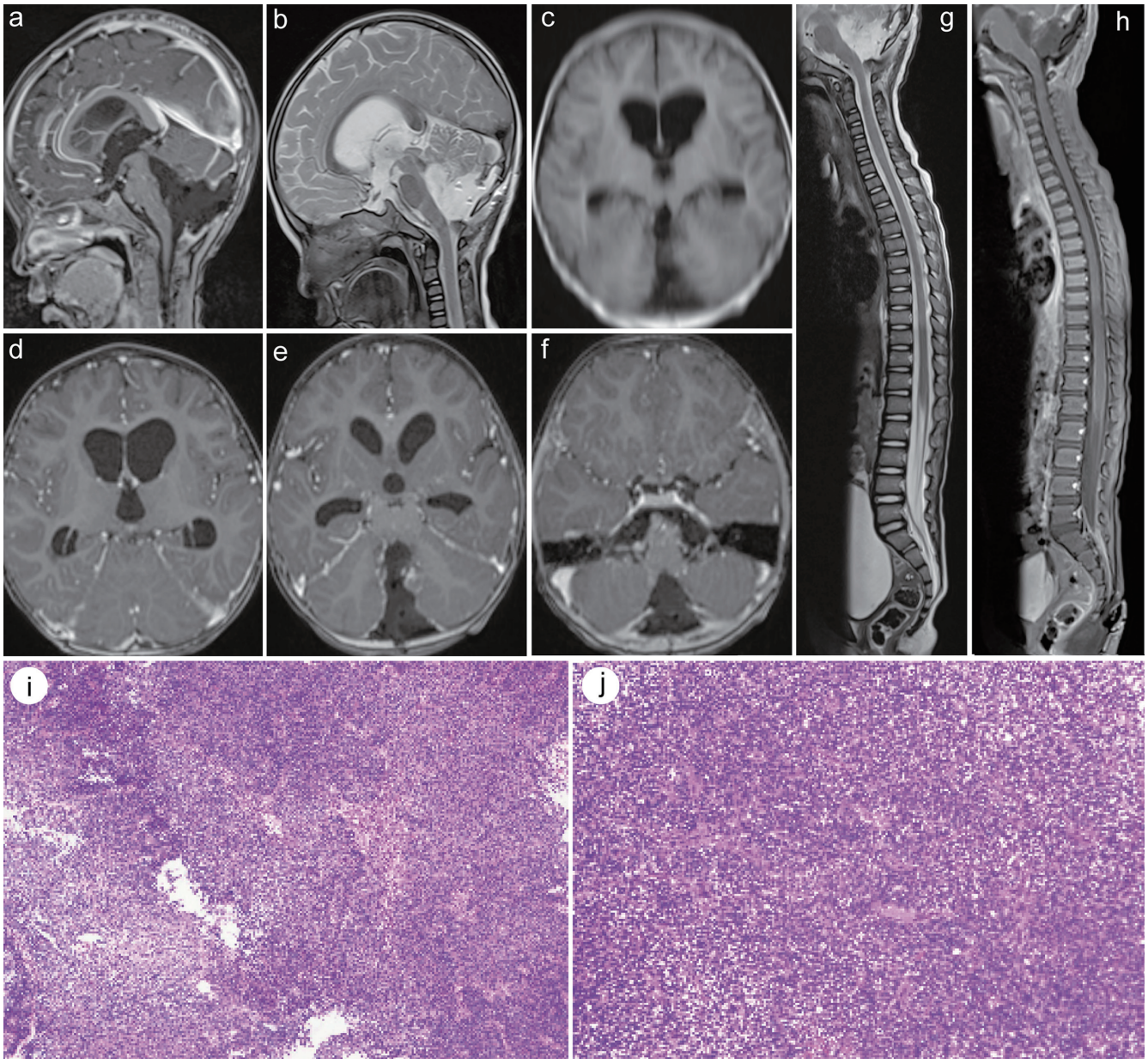

Fig. 2. Postoperative pathological and imaging manifestations of atypical teratoid/rhabdoid tumors. (a–f) The MRI in the sagittal and horizontal planes shows that the tumor has been completely removed. (g, h) The tumor did not metastasize to the distant spinal cord. (i, j) Postoperative pathological manifestations (200×). MRI, magnetic resonance imaging.

and scheduled follow-up visits.

Prognosis and survival

The patient was referred to the oncology department for adjuvant radiotherapy and chemotherapy, receiving a regimen that included vincristine and carboplatin. Despite aggressive multimodal therapy, the patient's quality of life remained poor, with persistent neurological deficits and limited functional recovery. The patient survived for eight months post-surgery but required frequent hospital readmissions due to complications, including infections and progressive neurological decline. This outcome highlights the aggressive nature of AT/RT and the limited effectiveness of current treatment modalities in improving long-term survival and quality of life.

Discussion

AT/RT represents one of the most challenging pediatric CNS malignancies due to its aggressive biology, resistance to conventional therapies, and dismal prognosis (median survival <12 months).[7] While it predominantly affects children under three years of age, its rarity (1–2% of pediatric brain tumors) further complicates standardized management.[2] To contextualize our findings within the existing literature, we systematically reviewed recent AT/RT

Table 1. Summary of similar case reports of atypical teratoid/rhabdoid tumor (AT/RT) in pediatric patients

Study	Age/Sex	Location	Symptoms	Treatment	Outcome	Key findings
Guo *et al*., 2022[8]	27 months/M	Cerebellum	Vomiting, ataxia	Subtotal resection, chemotherapy (vincristine, carboplatin)	Survived nine months, poor quality of life	Tumor recurrence leads to a poor prognosis
Shah *et al*., 2023[9]	10 months/F	Sellar	Polyuria, polydipsia, drowsiness	Biopsy surgery	Abandoning treatment	Early identification and timely multidisciplinary management are crucial
El Malih *et al*., 2023[10]	1 month/M	Cerebellum	Eye malformation	chemotherapy (vincristine and cyclophosphamide)	Survived one week	The importance of radiological imaging in evaluating this rare disease
Fujigasaki *et al*., 2012[11]	6 months/F	Fourth ventricle	Vomiting	Total resection, radiotherapy	Survived 26 years	Highlighted the importance of complete tumor resection
Syed *et al*., 2023[12]	15 months/F	Lumbar vertebra	Fussiness	Subtotal resection, chemotherapy, radiotherapy	Survived two years, no recurrence	Aggressive adjuvant therapy can allow patients to achieve remission
Sharma *et al*., 2020[13]	4 years/M	Lateral ventricle	Vomiting, headache	Gross total resection	Survived three months, recurrence at two months	AT/RT can have an aggressive course with a tendency for early leptomeningeal spread

F, female; M, male.

case reports focusing on cerebellar presentations.

We conducted a systematic literature search of the PubMed and Web of Science databases for case reports published between 2000 and 2023. The search terms included "atypical teratoid/rhabdoid tumor", "AT/RT", "pediatric", and "cerebellum". A total of 15 relevant case reports were identified, and 6 representative cases involving children under three years old were selected for inclusion based on their relevance to cerebellar AT/RT and the availability of detailed clinical data.

As summarized in Table 1, AT/RT is among the most malignant forms of childhood CNS neoplasms.[8–13] The selected reports provide a comprehensive overview of the clinical features, treatment strategies, and outcomes of pediatric AT/RT. They highlight the challenges in managing this aggressive tumor and underscore the urgent need for further research into targeted therapies and personalized treatment approaches.

Patients with AT/RT usually present within the first three years of life, with a slight male predominance.[2] In our case, the patient was a two-year-old girl, aligning with the typical age of onset but contrasting with the reported male predilection. Symptoms commonly include signs of increased intracranial pressure, such as vomiting, headaches, and papilledema, along with focal neurological deficits like seizures and motor or sensory impairments.[14] Our patient exhibited frequent vomiting and gait instability, consistent with cerebellar dysfunction due to the tumor's location in the fourth ventricle. Radiological imaging typically shows a heterogeneous mass with mixed density on computed tomography and variable signal intensity on T1-weighted and T2-weighted MRI images.[5] Post-contrast enhancement often reveals irregular patterns of tumor vascularity. In this case, preoperative MRI revealed a well-circumscribed cerebellar mass with mixed T1/T2 signals and post-contrast enhancement (Fig. 1a–c), consistent with classical imaging features.

Definitive diagnosis relies on surgical resection and histopathological examination. Distinguishing AT/RT from other pediatric brain tumors, particularly medulloblastoma and primitive neuroectodermal tumors, requires careful evaluation of immunohistochemical markers.[15] Genetic studies have identified specific alterations in AT/RT, with SMARCB1 deletion being a key event in its pathogenesis,[6] providing potential therapeutic targets.

Given the aggressive nature of AT/RT, multimodal therapy is recommended.[16] Our patient underwent a gross total resection followed by adjuvant radiotherapy and chemotherapy. Despite achieving radiological remission (Fig. 2a–f), survival was limited to eight months, with severe neurological deficits and recurrent infections. Adjuvant radiotherapy targets residual disease and has demonstrated some efficacy in delaying progression.[17] Chemotherapy regimens, including high-dose methotrexate, vincristine, and carboplatin, are commonly employed, but response rates remain suboptimal.[7] Targeted therapies, including enhancer of zeste homolog 2 inhibitors (e.g., UNC1999), mesenchymal-epithelial transition factor inhibitors, and immune checkpoint inhibitors, are currently under investigation in clinical trials and offer hope for improved survival outcomes.[6]

Despite aggressive treatment, our patient's poor quality of life and limited survival underscore the grim prognosis associated with AT/RT. The tumor's rapid progression and resistance to conventional therapies continue to pose significant therapeutic challenges.[6] A deeper understanding of the molecular mechanisms underlying this disease is crucial for the development of more effective treatments.

Conclusions

This case highlights the challenges associated with managing AT/RT, particularly in pediatric patients. Complete surgical resection remains the cornerstone of AT/RT treatment, as demonstrated in this case, where total tumor removal was achieved. However, despite successful surgery and multimodal therapy, the prognosis for AT/RT remains poor, underscoring the urgent need for more effective treatment strategies. Postoperative care and rehabilitation are

crucial for optimizing patient recovery and quality of life. Collaborative efforts among clinicians, pathologists, and researchers are essential to advancing our understanding of this devastating disease and to the development of innovative therapeutic approaches.

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Conflict of interest

The authors declare that they have no known competing financial interests or personal relationships that could have influenced the work reported in this paper.

Author contributions

Conceptualization, methodology, investigation of this study (XH, HL), data curation, formal analysis, writing the original draft (XH), validation, resources, and supervision (YKC). All authors participated in the review and editing of the manuscript and approved the final version for publication.

Ethical statement

This study was conducted in accordance with the ethical standards of Huazhong University of Science and Technology Tongji Medical College Medical Ethics Committee (TJ-IRB2025068), as well as the 2024 Helsinki Declaration and its later amendments. Informed consent was obtained from the patient's legal guardian for the publication of this case report and any accompanying images.

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