Clinical Case Report

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Atypical teratoid rhabdoid tumor mimicking type II neurofibromatosis

A case report

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Abstract

Rationale: Brain magnetic resonance imaging (MRI) images of atypical teratoid rhabdoid tumor (ATRT) often present heterogeneous signals of various cells without remarkable features of the disease. We describe a unique case of atypical brain MRI images presenting as an type II neurofibromatosis and explore some diagnostic hints.

Patient concerns: A 1-year-and-7-month-old boy admitted to our department with a 7-day history of drowsiness and 2-day history of emesis, and his presenting complaint was repeated vomit. On physical examination, he had drowsiness, positive sun set sign, slow light reflection, high muscular tension of limbs and 55 cm head circumference. MRI presented masses of bilateral auditory nerve distribution area, the fourth ventricle and right frontal lobe, obstructive hydrocephalus, and amplified cisterna magna. Particularly, dumbbell shape tumor in left cerebellopontine angle area and the fourth ventricle showed iso- or hypo-intensity on T1-weighted image and mix-intensity on T2-weighted image with irregular frontier, obvious mutual high and low signal on T2-weighted image, and growing along cerebrospinal fluid pathway.

Diagnosis: The diagnosis of type II neurofibromatosis (NF-II) was considered pre-operatively. After surgery, postoperative histopathology confirmed the diagnosis of ATRT.

Interventions: After ventriculo-peritoneal (VP) shunt, no evidence of tumor was inspected in cerebrospinal fluid, and enhancement MRI showed heterogeneous contrast signal on dumbbell shape tumor. We executed an incomplete microsurgery for dumbbell shape lesion in left auditory nerve distribution area and the fourth ventricle for differential diagnosis and facilitating further treatment.

Outcomes: The patient did not recover well postoperatively and suffered from severe pulmonary infection. Refusing further intervention in view of poor prognosis of ATRT, the patient was transferred to another hospital for rehabilitation care. The patient died from progressive tumor and respiratory failure after 2 months.

Lessons: The diagnosis of ATRT can be challenging, in our case due to the disturbance of bilateral auditory nerve distribution area tumors. Under MRI, Irregular frontier, obvious mutual high and low signal on T2-weighted image, growing along cerebrospinal fluid pathway, and heterogeneous contrast enhancement should lead the clinician to strongly consider ATRT.

Abbreviations: ATRT = atypical teratoid rhabdoid tumor, MRI = magnetic resonance imaging, NF-II = type II neurofibromatosis, VP = ventriculo-peritoneal.

Keywords: ATRT, MRI, NF-II

1. Introduction

Atypical teratoid rhabdoid tumor (ATRT) is a malignant central nervous system neoplasm primarily occurs in children who are younger than two years old.[1] Though a variety of therapies have been used in patients with ATRT, they have suffered a dismal outcome of rapid recurrence and death with median survival time reported less than one year.[2] In typical magnetic resonance imaging (MRI) of brain, T1 weighted image of ATRT shows iso- or hypo-intensity, and T2 weighted image shows iso- or hyper-intensity, with varied contrast enhancement after contrast-medium administration.[3] Herein, we described the case of a child who presented with atypical head MRI images, without remarkable typical features of the disease.

2. Case report

The Ethics Committee of the Children's Hospital, Zhejiang University School of Medicine approved the study (2019-IRB-002).

Informed written consent was obtained from the patient's family for publication of this case report and accompanying images.

A 1-year-and-7-month-old boy presented to the emergency room with a 7-day history of drowsiness and 2-day history of emesis. His parents are descendants from consanguineous marriage family (Fig. 1). On physical examination, he had

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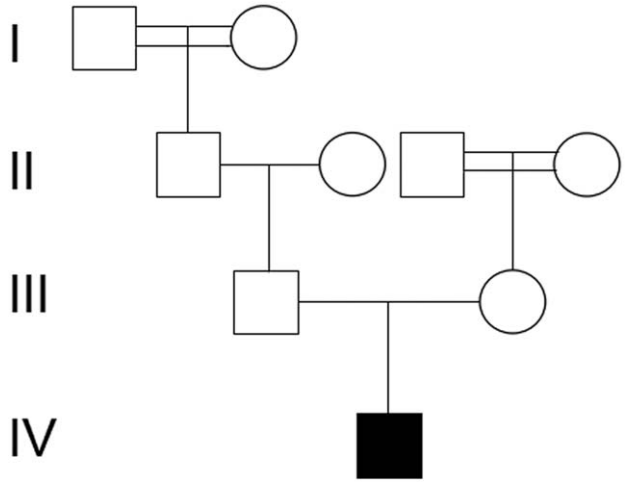

Figure 1. The patient's family: our patient was the fourth generation, and the first generation of patriarchal lineage and second generation of maternal lineage suffered from consanguineous marriage.

drowsiness, positive sun set sign, slow light reflection, high muscular tension of limbs and 55 cm head circumference. CT showed multiple nodules of bilateral bridge cerebellar angle and the fourth ventricle, arachnoid cyst of left middle cranial fossa and obstructive hydrocephalus. During the period of pre-operative preparation for ventriculoperitoneal VP shunt, an emergency MRI presented masses of bilateral auditory nerve distribution area, the fourth ventricle and right frontal lobe (Fig. 2A–D), obstructive hydrocephalus, and amplified cisterna magna (Fig. 2B and D). Particularly, dumbbell shape tumor in left cerebellopontine angle area and fourth ventricle showed iso- or hypo-intensity on T1-weighted image and mix-intensity on T2-weighted image with irregular frontier, obvious mutual high and low signal on T2-weighted image, and growing along cerebrospinal fluid pathway (Fig. 2A and B). After VP shunt, no evidence of tumor was found in cerebrospinal fluid, and MRI enhancement scanning showed no remarkable contrast enhancement in frontal lesion and heterogeneous contrast enhancement in the remaining lesions (Figs. 3A–D and 4A–C).

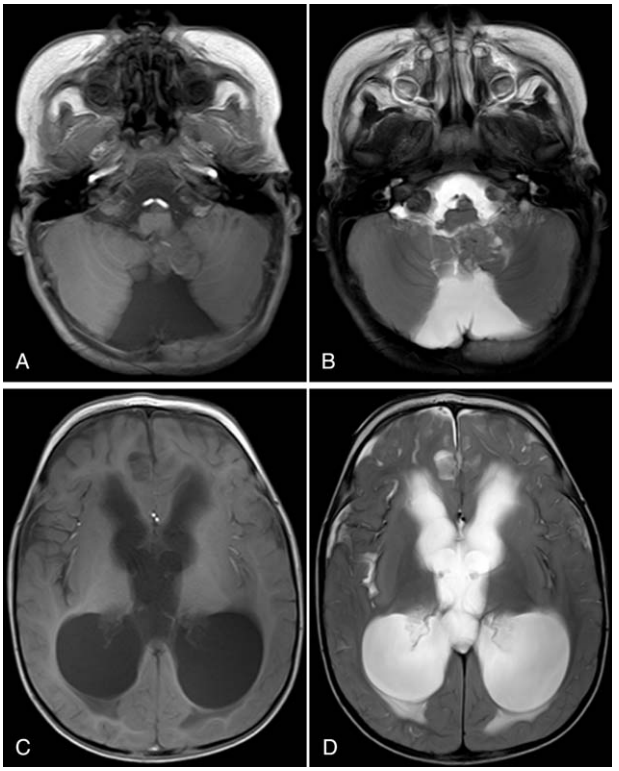

Figure 2. Head MRI: showed masses of bilateral auditory nerve distribution area, fourth ventricle and right frontal lobe, obstructive hydrocephalus, and amplified cisterna magna.

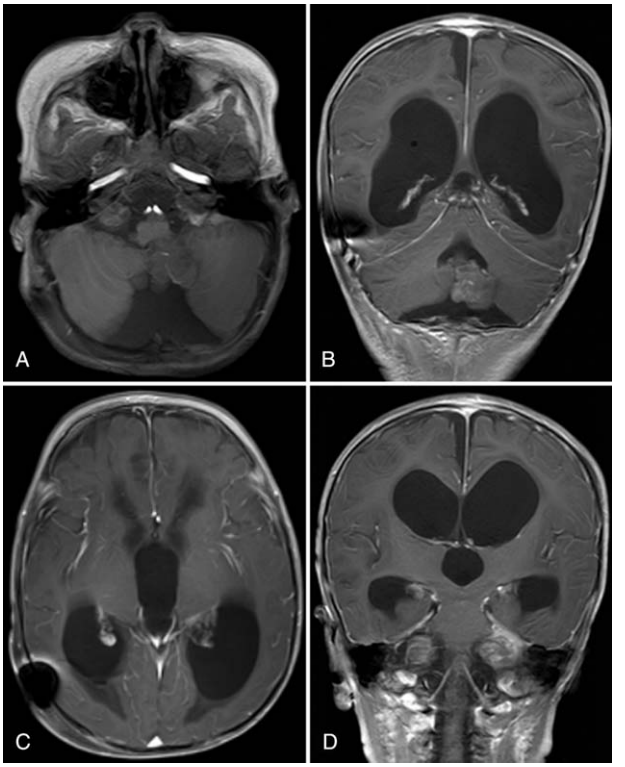

Figure 3. MR enhancement scanning: (A, C) axial view and (B, D) coronal view showed no remarkable contrast enhancement in frontal lesion and inhomogenous contrast enhancement in the lesions of posterior cranial fossa.

In order to rule out the diagnosis of NF-II and to facilitate further treatment, we executed a microsurgery for dumbbell-like lesions in posterior cranial fossa. Histopathology confined the diagnosis of ATRT. There were confluent oval or spindle shaped cells with obvious nucleolus and vacuolization in parts of them, and abundant undifferentiated cells under microscope (Fig. 5A), and immunohistochemistry staining showed INI1 (Fig. 5B) and OLIG2 were negative, GFAP and S-100 were positive, EMA, CK, and Syn were weakly positive, and 75% positive Ki-67 was positive. Genetic sequencing presented MSH3-p.Ala61_Pro63dup mutation without a targeted drug. Unfortunately, the patient suffered severe pulmonary infection postoperatively and did not recover well. Refusing further intervention in view of poor prognosis of ATRT, the patient was transferred to another hospital for rehabilitation care. The patient died from progressive tumor and respiratory failure after 2 months.

3. Discussion

ATRT is a rare malignant disease and the exact incidence rate of ATRT is difficult to determine, since it has been recognized in

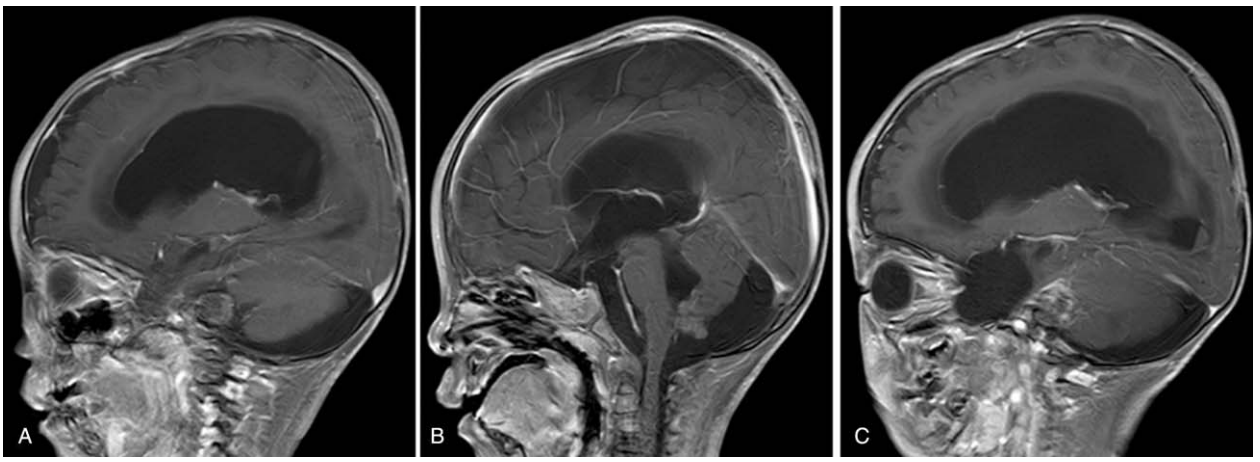

Figure 4. MR enhancement scanning: (A–C) sagittal view showed no remarkable contrast enhancement in frontal lesion and inhomogenous contrast enhancement in the lesions of posterior cranial fossa.

1996.[4] Most published data concludes poor outcome of ATRT, however, reports of long-term survivors exist.[5,6] This disease has no characteristic MRI presentation and its diagnosis relies on pathology. Immunostaining for loss of SMARCB1 protein expression helps to confirm the diagnosis.[7] Therefore, we report a ATRT case with atypical NF-II–like MRI images.

NF-II is an autosomal dominant disorder and characterized by multiple tumors involving the central nervous system. The incidence of NF-II is about 1 in 40,000 individuals.[8] Commonly MRI presents bilateral vestibular schwannoma, located in the internal auditory canal and often extended into the cerebellopontine angle and the typical "ice cream cone" appearance can be seen.[9] These lesions are hypointense on T1-weighted images and hyperintense on T2-weighted images under intense contrast enhancement.

We report the case of a 1-year-and-7-month-old boy with a 7-day history of drowsiness and 2-day history of emesis. His brain MRI was characteristic of multiple brain tumors, masses in bilateral auditory nerve distribution area, which misled us with diagnosis of NF-II. However, there were other characters of irregular frontier, obvious mutual high and low signal on T2-weighted image, tumor growing along cerebrospinal fluid pathway, and heterogeneous contrast enhancement. Eventually, the histopathology confirmed the diagnosis of ATRT. We speculated above characters were conducive to diagnostic evaluation of ATRT.

Images of reported ATRTs have no definite specificity. In 2008, Warmuth-Metz et al[10] reported a rather unusual pattern of contrast enhancement which may be typical for ATRTs. Arslanoglu et al[11] recommended solid-cystic mass in central

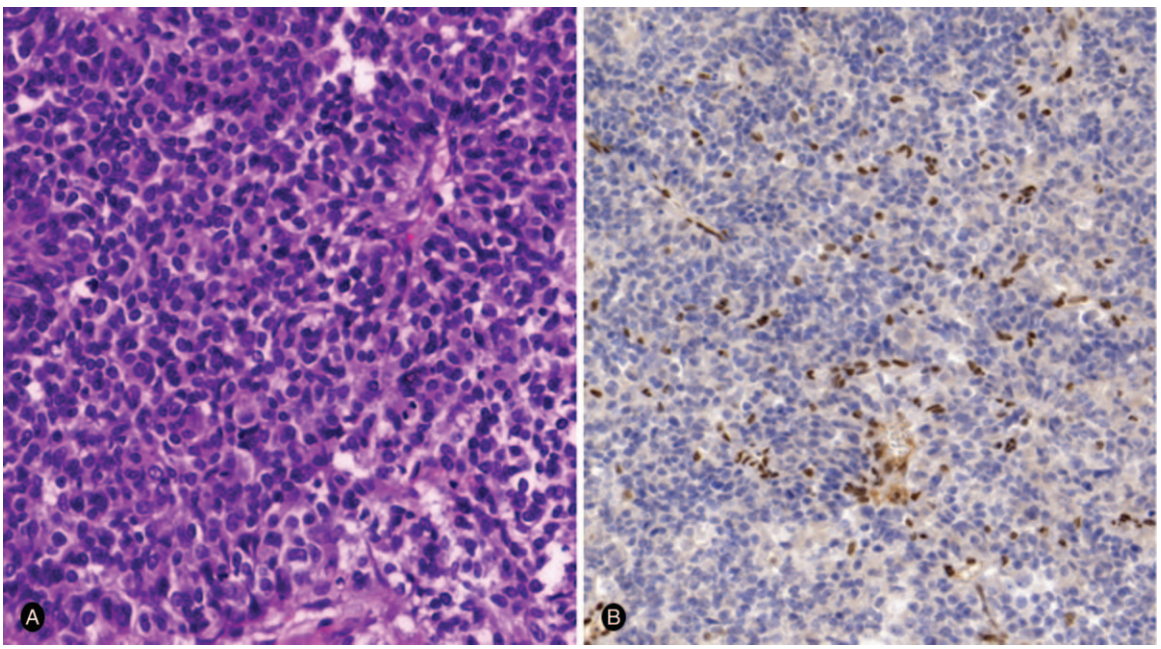

Figure 5. (A) H-E staining showed confluent oval or spindle shaped cells with obvious nucleolus and vacuolization in parts of them, and abundant undifferentiated cells (40×). (B) Immunohistochemistry showed negative INI1.

line of posterior cranial fossa with hemorrhage, necrosis, calcification and the patients younger than 2 years old as hints of ATRT. Most often, MRI shows heterogeneous signals of various cells, hemorrhage, necrosis, and calcification.[10] And the tumor has different degrees of enhancement after contrast-medium administration. From our case, we found characteristics, especially the obvious mutual high and low signal on T2-weighted image and tumor growing along cerebrospinal fluid pathway, due to ATRT's characteristic to disseminate along cerebrospinal fluid pathway, may be specific manifestations of MRI for ATRT.

Though MRI is widely used, the diagnosis of ATRT still depends on histopathology examination. INI1 is the most important marker of immunohistochemistry, and it is negative in approximately 75% of ATRT.[12] Treatment strategy is commonly combined multiple therapies including surgery, radiotherapy, and chemotherapy, ATRT had poor outcome though. Recently, Ren et al reported that the 2-year overall survival rate and event-free survival rate for 18 consecutive patients were 33.3% and 27.8%, respectively.[13] Treatments performed were as follows: surgery alone in two patients, surgery and radiation therapy in two patients, surgery and chemotherapy in five patients, surgery combined with chemotherapy and radiation therapy in two patients, and surgery combined with chemotherapy, radiation therapy, and gamma knife surgery in seven patients.

To sum up, the diagnosis of ATRT can be challenging because of a lack of specific image signs. Irregular frontier, obvious mutual high and low signal on T2-weighted image, growing along cerebrospinal fluid pathway, and heterogeneous contrast enhancement should lead the clinician to strongly consider the diagnosis of ATRT.

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Validation: Hanhai Zeng.
Visualization: Hanhai Zeng.
Writing – original draft: Hanhai Zeng.
Writing – review & editing: Hanhai Zeng.

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