

Case Report

Atypical teratoid rhabdoid tumor (AT/RT) at the cerebellopontine angle arising from acoustic nerve with facial nerve palsy: report of a case

Pao-Shu Wu^{1,2}

¹Department of Pathology, MacKay Memorial Hospital, ²Mackay Junior College of Medicine, Nursing, Management, Taipei, Taiwan

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Abstract: Atypical teratoid rhabdoid tumor (AT/RT) is a highly aggressive central nervous system embryonic tumor predominantly occurring in infants and young children. The AT/RT can occur in the cerebellopontine angle and cause facial nerve palsy as a presenting sign. We present a case of a 2-year-old girl with a cerebellopontine angle tumor who initially presented with acute facial palsy. Later, a subsequent diagnosis of AT/RT was made.

Keywords: Atypical teratoid rhabdoid tumor, cerebellopontine angle, acoustic nerve, facial nerve palsy

Introduction

Atypical teratoid rhabdoid tumors (AT/RTs) are highly malignant central nervous system (CNS) neoplasm predominantly occurring in infants or young children younger than 3 years. AT/RTs are a type of embryonic CNS tumor demonstrating histologic and immunohistochemical evidence of polyphenotypic differentiation with neuroectodermal, epithelial, and mesenchymal lines [1]. AT/RT corresponds histologically to WHO grade IV neoplasms that carry a poor prognosis with a 3-year overall survival rate of 22% [1, 2]. AT/RTs can occur in both supra- and infra-tentorial regions including cerebral hemispheres, ventricular system, suprasellar region, pineal gland, cerebellar hemispheres, brain stem, and cerebellopontine angle (CP angle) [3, 4]. With AT/RT in the CP angle it is relatively difficult for cytoreductive surgery to decrease tumor burden, because the CP angle contains many critical structures [5]. Moreover, a more common and frequent lesion in CP angle, schwannoma, might be assumed and would delay treatment. Thus, preoperative differential diagnosis is important for surgical planning and prognostication. Here we report a case of a 2-year-old girl who presented with facial nerve palsy. Subsequent diagnosis was a CP angle AT/RT.

Case report

A 2-year-old girl presented with acute left facial palsy and hearing loss 4 months before visiting our hospital with an initial diagnosis of viral infection. Administration of anti-viral agents and steroids did not relieve the symptoms. In our hospital, brain computed tomography (CT) and magnetic resonance imaging (MRI) examinations were performed and the results revealed a 1.6 × 1.5 cm solid mass in the left CP angle with widened internal auditory meatus (**Figure 1A, 1B**). The initial radiological impression was benign schwannoma arising from acoustic nerve. She underwent a craniectomy to obtain a biopsy specimen of the lesion.

Histopathologically, the tumor showed high cellularity and was composed of primitive small blue round cells with abundant eosinophilic cytoplasm, eccentrically located nuclei containing vesicular chromatin, and conspicuous eosinophilic nucleoli (**Figure 2A**). Immunohistochemically, tumor cells were strongly diffuse positive for vimentin, and focally positive for smooth muscle actin (SMA) (**Figure 2B, 2C**), while negative for lymphoma marker CD45, rhabdomyosarcoma marker MyoD1, and Ewing sarcoma/PNET (primitive neuroectodermal tumor) marker NKX2.2 (not shown). The

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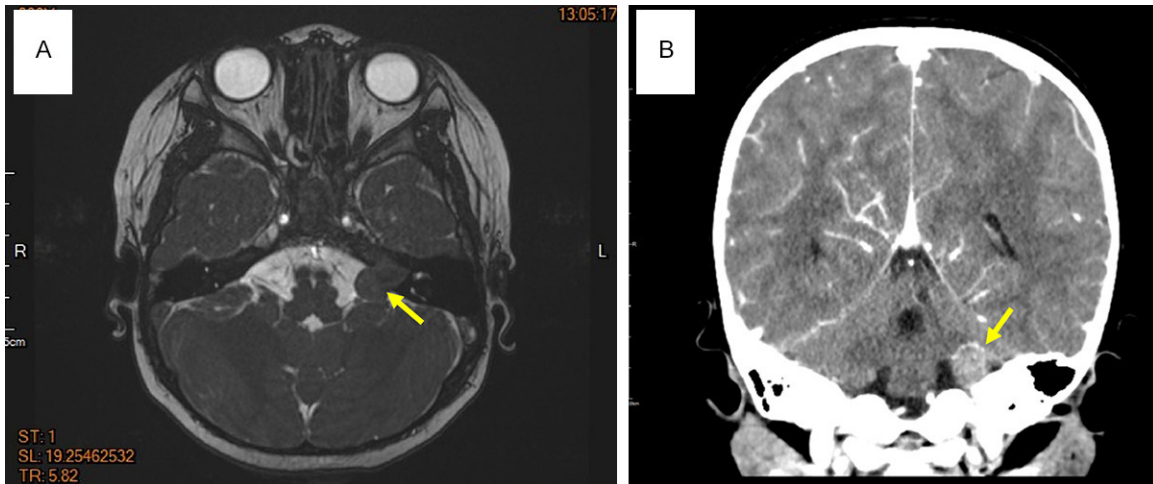


Figure 1. Brain MRI and CT images showed a 1.6 × 1.5 cm heterogeneously enhancing tumor in the left CP angle cistern with extension into the left internal acoustic canal. (A) Transverse view (MRI) (yellow arrow), and (B) coronal view (CT) (yellow arrow).

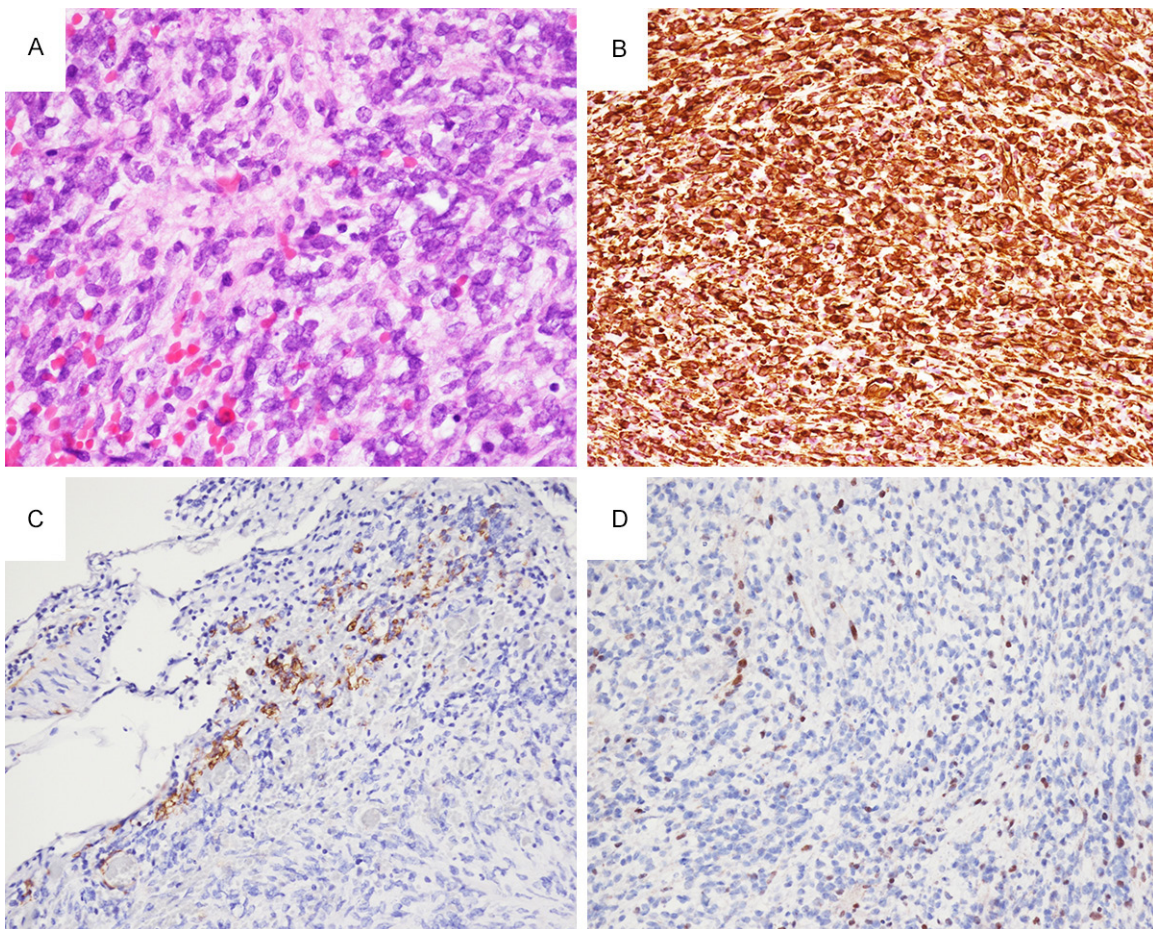


Figure 2. (A) Hematoxylin and eosin stain of the tumor showed primitive small round to rhabdoid cells with abundant eosinophilic cytoplasm, vesicular chromatin, and conspicuous eosinophilic nucleoli. Tumor cells strongly express vimentin (B), and focally express smooth muscle actin (C). (D) Loss of expression of INI1/SMARCB1 in nuclei of tumor cells, but with retained expression in intratumoral lymphocytes and endothelial cells of blood vessel (200 ×).

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Ki-67 proliferation index was high at about 45% (not shown). Pathognomonic immunostain was the complete loss of INI1/SMARCB1 in tumor cells (**Figure 2D**), supporting the diagnosis of AT/RT.

The patient recovered well from the surgery and was transferred to pediatric oncology department for the subsequent chemotherapy plan. Further physical survey revealed the dissemination of AT/RT tumor cells in cerebrospinal fluid. She subsequently received systemic chemotherapy including cisplatin and vincristine regime and was in good condition one year after the surgery.

Discussion

AT/RTs account for approximately 1-2% of pediatric brain tumors but are estimated to account for > 10% of CNS tumors in infants due to the preponderance of cases in children < 3 years [6, 7]. On the other hand, cerebellopontine angle tumors in young children are infrequent and accounting for less than 3% of pediatric brain tumors [8]. The clinical presentation of AT/RT is variable and depending on the age of the patient and the size and location of the tumor. Infants are more often present with non-specific signs of vomiting, lethargy or failure to thrive. More specific signs include head tilt and cranial nerve palsy, most commonly sixth and seventh paresis [1]. A series study showed up to 46% of patients with AT/RT present with cranial nerve palsies as the initial symptom [9]. The nature of AT/RT as it relates to specific cranial nerves such as facial nerve is unknown.

The exact histogenesis of AT/RT is not well understood. Although the cell of origin in AT/RTs is unclear, it has been suggested that they derive from pluripotent fetal cells since neural, epithelial, and mesenchymal markers can all be expressed [10, 11]. AT/RTs can occur sporadically or as part of a rhabdoid tumor predisposition syndrome [6]. Mutation or loss of *INI1/SMARCB1* locus at 22q11.2 is the genetic hallmark of AT/RT [12]. The INI1/SMARCB1 protein is one component of the mammalian SWI/SNF complex, which functions as an ATP-dependent chromatin remodeler and regulates a plethora of gene expressions [13]. Loss of INI1/SMARCB1 results in widespread but specific deregulation of genes and pathways associated with cell cycle, cell survival, differentiation and the epigenome [14, 15].

The differential diagnosis in young children with CP angle tumors, according to several large case series, commonly includes medulloblastoma, schwannoma, ependymoma, and astrocytoma [8, 16, 17]. However, no specific symptoms are correlated to pathologic findings on the above studies. Siu et al. reported that in three cases, acute facial nerve palsy is highly associated with CP angle AT/RT, indicating acute facial nerve neuropathy may serve as an adjunct that should raise the suspicion for AT/RT in CP angle lesions in children [5]. Albeit rare, clinicians and pathologists should be aware of the acuity of facial nerve deficits as this may raise the concern for AT/RT in the pre-operative differential diagnosis and could guide the treatment course and prompt implementation of a multimodal therapy plan as early as possible.

Disclosure of conflict of interest

None.

Address correspondence to: Dr. Pao-Shu Wu, Department of Pathology, MacKay Memorial Hospital, No.45, Minsheng Rd., Tamsui District, New Taipei City 251, Taiwan. Tel: +886-2-28094661; Fax: +886-2-28093385; E-mail: pw2136@gmail.com

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