Atypical Teratoid Rhabdoid Tumor in a Child: Case Report

Tumor teratóide rabdóide atípico em uma criança: relato de caso

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Arq Bras Neurocir 2015;34:241–244.

Abstract

Atypical teratoid/rhabdoid tumor (ATRT) is a rare and aggressive type of embryonal tumor of the central nervous system (CNS) occurring in childhood. The present study aims to describe the case of a 16-year-old girl who presented with an occipital mass lesion that was diagnosed as ATRT. We present a brief review of the current knowledge of the treatment of this rare neoplasm. A previously healthy 16-year-old girl was referred after two episodes of partial complex seizure 2 weeks before admission. MRI showed a right parieto-occipital lesion with homogeneous contrast-enhancing and significant surrounding brain edema. The patient underwent uneventful surgical resection of the lesion and was discharged home on the fifth postoperative day. Pathologic examination revealed it to be ATRT. The patient was referred to chemotherapy and radiotherapy. After 6 months of follow-up, the patient remains free of seizure and disease progression. ATRT is a rare and aggressive disease. Therefore, early diagnosis and treatment may improve the patient's prognosis and quality of life.

Keywords
- atypical teratoid/rhabdoid tumor
- childhood
- embryonal tumor

Resumo

O tumor teratoide rabdoide atípico (TTRA) é um tipo de neoplasia rara, com comportamento maligno, que atinge o sistema nervoso central (SNC) de crianças. O objetivo do presente estudo é relatar o caso de uma paciente de 16 anos de idade admitida no nosso serviço com uma lesão occipital que revelou tratar-se de TTRA em estudo anatomopatológico. Uma paciente previamente hígida foi admitida no nosso serviço referindo ocorrência de dois episódios de crises convulsivas nas últimas 2 semanas. A ressonância magnética de crânio mostrou a presença de lesão occipital direita com captação homogênea do meio de contraste. A paciente foi submetida a cirurgia de ressecção da lesão expansiva sem intercorrências. A análise histopatológica

Palavras-chave
- tumor teratoide rabdoide atípico (TTRA)
- criança
- tumor embrionário

received February 4, 2015
accepted June 12, 2015

DOI http://dx.doi.org/10.1055/s-0035-1560034.
ISSN 0103-5355.

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e imuno-histoquímica revelou tratar-se de TTRA. A paciente foi encaminhada para a realização de quimioterapia e radioterapia complementares ao tratamento cirúrgico. Após 6 meses de seguimento ambulatorial, a paciente encontra-se assintomática, sem recorrência das crises convulsivas e/ou progressão radiológica da doença. TTRA é uma doença rara e agressiva. Com isso, diagnóstico e tratamento antecipados podem aprimorar prognóstico e qualidade de vida dos pacientes.

Introduction

Atypical teratoid/rhabdoid tumor (ATRT) is a rare and aggressive type of embryonal tumor of the central nervous system (CNS) occurring in childhood.[1–3] The incidence of all embryonal tumors in the United States is 0.66/100,000 in persons 0 to 19 years old.[4] ATRT commonly affects infants and young children, especially children younger than 3 years, and prognosis is generally poor.[5] There is no standard treatment regimen for these tumors. Complete surgical resection is usually a challenge due to tumor location in the brain and young age at diagnosis. Additionally, radiation is often avoided in very young children due to severe neurocognitive late effects.[6] Intensive chemotherapy regimens are currently under study, and no standard chemotherapy regimen exists for these patients.[7]

This study aims to describe the case of a 16-year-old girl who presented with an occipital mass lesion that was diagnosed as ATRT. We present a brief review of the current knowledge of the treatment of this rare neoplasm.

Case Report

A previously healthy 16-year-old girl was referred after two episodes of partial complex seizure 2 weeks before admission. Magnetic resonance imaging (MRI) showed a right parieto-occipital lesion with homogeneous contrast-enhancing (►**Fig. 1**) and significant surrounding brain edema. The patient underwent uneventful surgical resection of the lesion and was discharged home on the fifth postoperative day. Microscopically, a tumor containing primitive neuroectodermal and rhabdoid cells was detected (►**Fig. 2**). The rhabdoid component was composed of medium-sized, round to oval cells with eccentric nuclei, prominent nucleoli, and eosinophilic cytoplasm. Immunohistologically, the rhabdoid cells were immunoreactive with smooth muscle actin (SMA), epithelial membrane antigen (EMA), and vimentin, but negative with desmin. The small cell embryonal component expressed vimentin. Ki67 was detected to be 15%. The morphologic features were compatible with diagnosis of ATRT (►**Fig. 3**). The patient was referred to chemotherapy and radiotherapy. After 6 months of follow-up, the patient remains free of seizure and disease progression.

This case report was analyzed and approved by the Ethical Committee of our institution.

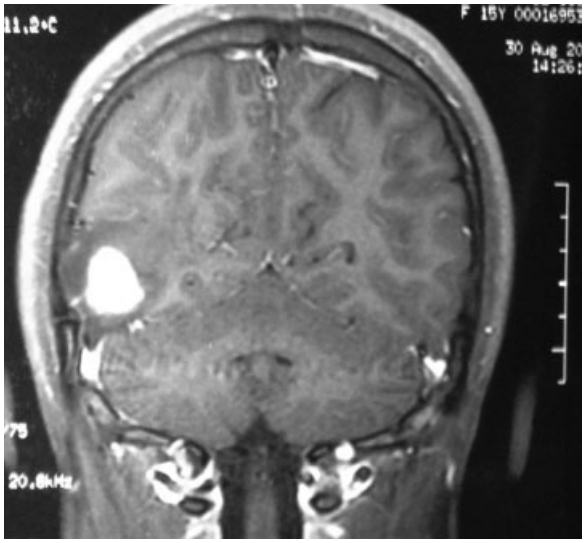

Fig. 1 MRI showing contrast enhancing right parieto-occipital nodular lesion.

Discussion

ATRT was first described as a distinct of CNS rhabdoid tumor in 1987 and was further described in 1996.[8] The updated World Health Organization (WHO) classification of CNS tumors in 2000 included ATRT for the first time.[9] ATRT belongs to a wider group of malignant rhabdoid tumors, which also include rhabdoid tumors of the kidney and extrarenal rhabdoid, which may develop in the lung, liver, and soft tissues.[10,11] Common features of malignant rhabdoid tumors are inactivation of *INI1* (also called *hSNF5/INI1/SMARCB1/BAF47*) gene located in the chromosome 22q11.2,[12] presence of rhabdoid cells, and unique immunohistochemistry results such as presence of vimentin, epithelial membrane antigen, smooth muscle actin, and glial fibrillary.[13]

The definitive diagnosis of ATRT relies on pathologic examination. However, clinical features of intracranial hypertension and some radiologic characteristics may help establish an adequate diagnosis. Although any part of the CNS may be involved, ATRT is mainly found in the posterior fossa.[8,14,15] The left hemisphere is more prominently affected in older children and adults, and tumors may be located within or extending into the ventricles.[16–18] Imaging features usually reveal large lesions, with precontrast computed tomography (CT) of an iso- or slightly hyperdense mass in relation to gray matter, with associated parenchymal edema calcification and hemorrhage.[17–19] MRI findings usually show mixed signal intensity on T1- and T2-weighted images because of extensive

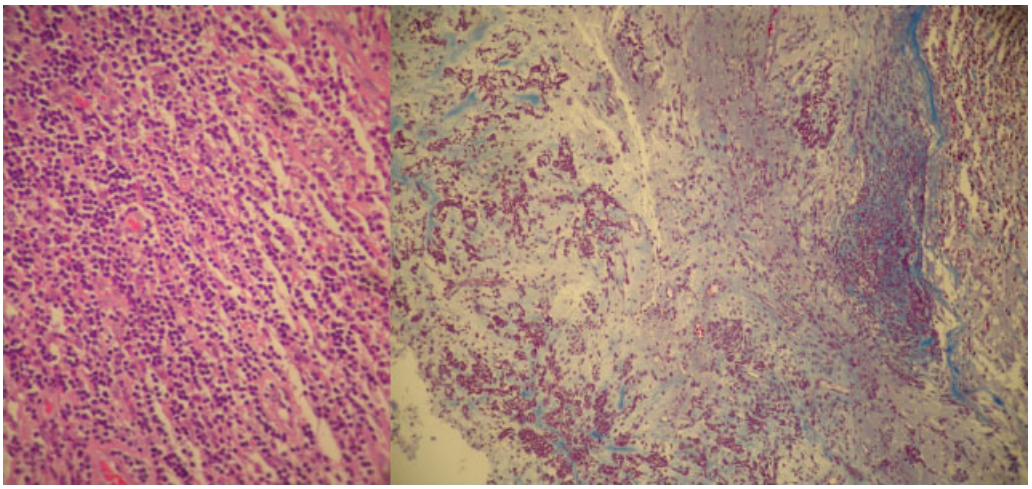

Fig. 2 Microscopic aspect of the tumor with rhabdoid eosinophilic cells and PNET-like cells.

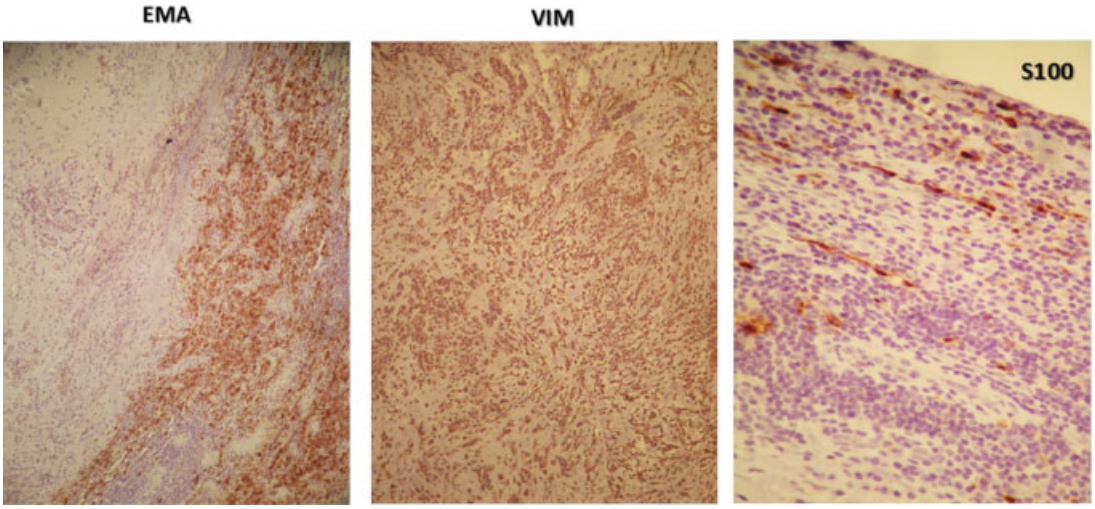

Fig. 3 Immunohistochemical study confirmed the characteristics of ATRT. Abbreviations: ATRT, atypical teratoid/rhabdoid tumor; EMA, epithelial membrane antigen.

necrosis and intratumoral hemorrhage.[17,20] The solid component of the tumor is hypo- or isointense on T1-weighted images and iso- or hyperintense on T2-weighted images compared with gray matter and is also well enhanced on postcontrast CT and MRI.[17–19,21]

Surgical treatment of ATRT has been proved as a favorable prognostic factor depending on the extension of resection. Gross total resection expanded median survival from 15 months (partial resection/biopsy) to 20 months.[5] Chemotherapy and radiotherapy also demonstrated favorable survival outcome in patients with ATRT; however, these therapies have been used with limitation due to the adverse effects and high risk of neurocognitive impairment in patients younger than 3 years.[5,12] In the present study, the patient was submitted to partial resection followed with radio- and chemotherapy. Our patient remains free of disease progression 6 months after surgery.

Conclusion

In conclusion, the present case highlights that ATRT is a rare and aggressive disease. Therefore, early diagnosis and treatment may improve the patient's prognosis and quality of life.

Consent Statement

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the editor of this journal.

Conflict of Interest

The authors declare that they have no competing interests.

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