Atypical Teratoid Rhabdoid Tumor in a Child: Case Report

Tumor teratóide rabdóide atípico em uma criança: relato de caso

Lucas Crociati Meguins¹ Ronaldo Brasileiro de Miranda Batista Fernandes¹ Adriana Giubilei Pimenta¹ Ricardo Lourenço Caramanti¹ Eliane Milharcix Zanovelo² Carmen Lúcia Penteado Lancellotti² Mário José Góes¹

¹Departamento de Ciências Neurológicas. Hospital de Base da Faculdade de Medicina de São José do Rio Preto (FAMERP), São José do Rio Preto, São Paulo, Brazil
²Departamento de Patologia, Hospital de Base da Faculdade de Medicina de São José do Rio Preto (FAMERP), São José do Rio Preto, São Paulo, Brazil

Address for correspondence Lucas Crociati Meguins, MD, Hospital de Base da Faculdade de Medicina de São José do Rio Preto (FAMERP), Rua Pedro Palotta, 101/31B. São José do Rio Preto, São Paulo, Brazil CEP 15092-205 (e-mail: lucascrociati@hotmail.com).

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

Palavras-chave
► tumor teratoide rhabdoide atípico (TTRA)
► criança
► tumor embrionário

Received
February 4, 2015
Accepted
June 12, 2015

ISSN 0103-5355.

Copyright © 2015 by Thieme Publicações Ltda, Rio de Janeiro, Brazil

License terms
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 (\(\text{\textbullet Fig. 1}\)) 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 (\(\text{\textbullet Fig. 2}\)). 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 (\(\text{\textbullet Fig. 3}\)). 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.

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.\(^8\)–\(^15\) 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. 1** MRI showing contrast enhancing right parieto-occipital nodular lesion.
necrosis and intratumoral hemorrhage.\textsuperscript{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.\textsuperscript{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.\textsuperscript{5} Chemo-therapy 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.\textsuperscript{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.
References


