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Case Report

Primary atypical teratoid rhabdoid tumor in the adult spine

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Abstract

Background: Atypical teratoid/rhabdoid tumor (ATRT) is a highly aggressive tumor of the central nervous system (WHO grade IV), which is most frequently found intracranially in young children and infants. Only three prior cases of primary ATRT involving the adult spine were found following a literature review, and the average survival for these patients was only 20 postoperative months.

Case Description: A 43 year-old female presented with an acute exacerbation of chronic neck pain. While awaiting magnetic resonance (MR) studies of the cervical spine, she was found pulseless in her room. Although cardiopulmonary resuscitation was successful, she was found to be quadriplegic. The subsequent cervical MR imaging revealed a C1-3 intradural, extramedullary ventrolateral mass, markedly compressing the upper cervical spinal cord. Following successful surgical resection of the lesion, which proved pathologically to be an ATRT, she was treated with a full course of fractionated radiation therapy. Over the successive 6-month period, her neurological examination continued to improve to 4-/5 functional strength in her upper extremities, however, remained with 2/5 nonfunctional strength in her legs.

Conclusions: ATRT involving the adult spine are rare and may often be misdiagnosed. This study points out that aggressive surgery followed by radiation therapy may improve outcome.

Key Words: ATRT tumor, atypical teratoid rhabdoid tumor, cervical laminectomy, cervical spine surgery, INI1, neurosurgery

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INTRODUCTION

Primary atypical teratoid rhabdoid tumor (ATRT) rarely involves the adult spine. We present the case of a 43-year-old female with an ATRT found at the C1-3 level of the cervical spine. It was intradural/extramedullary in location and was completely excised after performing multilevel laminectomies. The patient continued to do well for 6 months following surgery and radiation therapy. Here, we review the three other adult spinal cases of ATRT reported in the literature, and discussed their average survival of only 20 months.

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CASE DESCRIPTION

Presentation and surgery

A 43-year-old female with a significant history of chronic neck pain, migraine, and coronary artery disease presented acutely with marked exacerbation of chronic neck pain and headache. When initially seen, she was neurologically intact. While awaiting magnetic resonance imaging (MRI) studies of the brain and cervical spine, she was found unresponsive and pulseless and required full cardiopulmonary resuscitation. The cervical MRI documented a C1-3 intradural, extramedullary, ventrolateral mass nearly filling the spinal canal resulting in severe compression of the upper cervical cord. Marked cord edema, seen on the T2 image, was present from C5 through the medulla [Figure 1]. Following resuscitation, the patient was awake and alert, but was found to be quadriplegic, exhibiting no movement of sensation in all four extremities. She was immediately placed on intravenous steroids and taken to the operating room for a C1-3 decompressive laminectomy and gross total resection of the tumor [Figure 2]. The pathological diagnosis proved to be consistent with an ATRT [Figures 3-5].

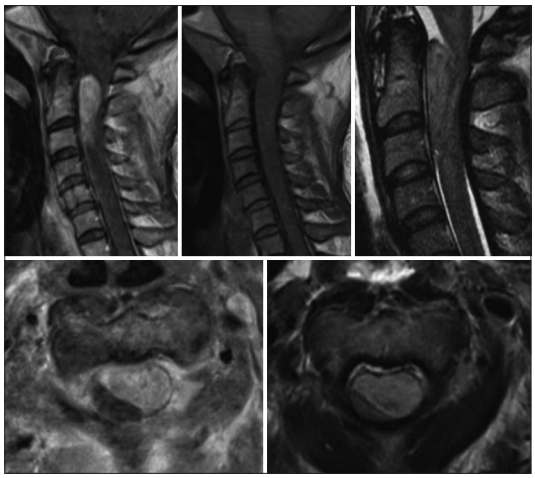

Figure 1: Pre-operative MRI. Clockwise from top left: T1 sagittal with contrast, T1 sagittal without contrast, T2 sagittal, T2 axial, T1 axial with contrast

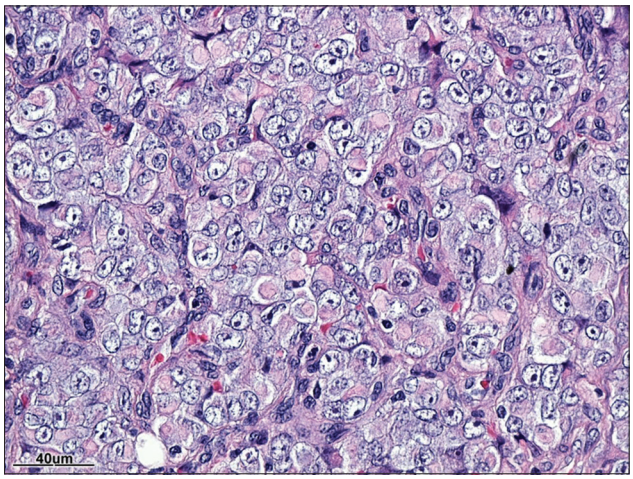

Figure 3: H and E stain. Sheets of polygonal rhabdoid tumor cells separated by an anastomosing capillary network. The rhabdoid cell is characterized with a large eccentric vesicular nucleus with a prominent nucleolus containing a round cytoplasmic eosinophilic inclusion

Postoperative course

Immediately postoperatively, the patient remained quadriplegic. However, on the postoperative day 12, she began moving the digits of the right hand and toes on the right side. She continued to exhibit poor respiratory effort and remained intubated for 14 days, subsequently requiring tracheostomy and gastrostomy. After 36 days of inpatient care, the patient was discharge to an inpatient rehabilitation facility. Upon discharge, she began a course of 28 fractions of radiation therapy for a total dose of 5040 cGy; she finished the radiation therapy 7 weeks

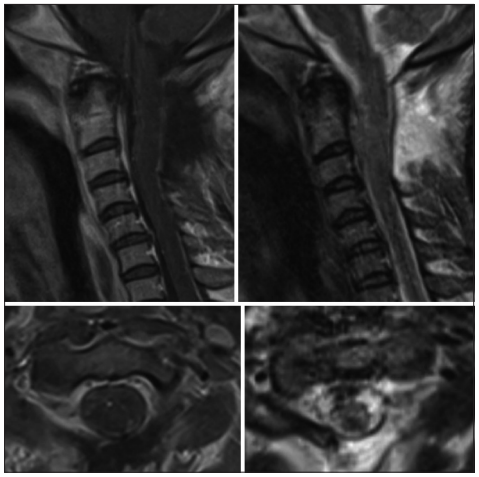

Figure 2: Postoperative MRI. Clockwise from top left: T1 sagittal with contrast, T2 sagittal, T2 axial, T1 axial with contrast

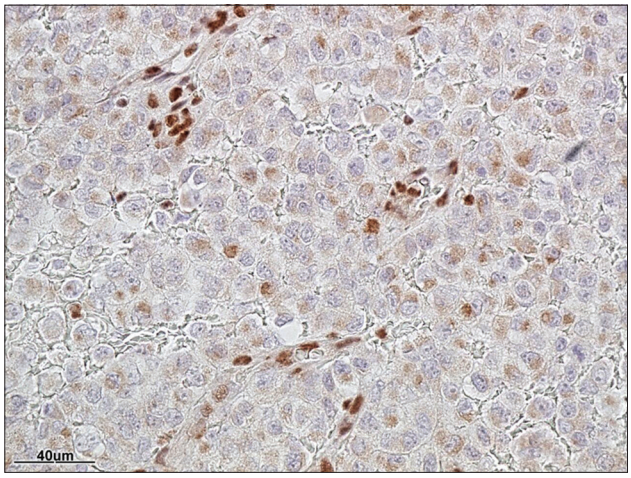

Figure 4: INI staining. Immunohistochemical stain for INI1 showing abnormal loss of nuclear staining of the rhabdoid cells with normal nuclear staining of the capillary endothelial cells and infiltrating lymphocytes. Very faint staining of the cytoplasmic inclusion is noted

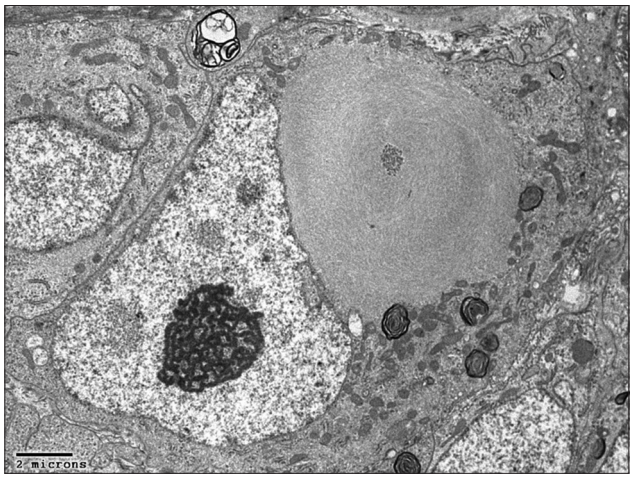

Figure 5: Electron microscopy. EM picture of a rhabdoid cell showing the vesicular nucleus with a large nucleolus and a pale whorled aggregate of intermediate filament displacing the formed cytoplasmic organelles

later. Because of low performance status, she was no deemed a suitable candidate for chemotherapy.

Twelve weeks postoperatively, she had regained antigravity strength in both the upper extremities and was able to feed herself. She also demonstrated gross movement in both the lower extremities in all muscle groups, however, this was still deemed nonfunctional strength. The subsequent cervical MRI at four months postopertive revealed no recurrent disease. At 6 months, the patient remained in a nursing home with a stable neurological exam.

Pathology

[Figures 3 and 5] Histopathological examination of the intradural, extramedullary, cervical spine tumor showed sheets of polygonal cells separated into small groups by a network of endothelial cell-lined capillaries. The tumor cells showed the characteristic rhabdoid appearance with an eccentric vesicular nucleus, prominent nucleolus, and the typical eosinophilic cytoplasmic inclusion. Electron microscopic examination of rhabdoid cells showed whorled aggregates of intermediate filaments making up the pink cytoplasmic inclusions seen in the hematoxylin and eosin (H and E) section. Immunohistochemical analysis depicted the typical immunophenotype of ATRT with loss of the nuclear INI1 (SMARCB1) staining, whereas staining positive for epithelial muscle antigen (EMA), vimentin, and smooth muscle antigen. In addition, the expected focal INI1 staining was positive in the surrounding endothelial cells and infiltrating leukocytes. The final diagnosis was ATRT [Figures 1 and 5].

DISCUSSION

ATRT are highly aggressive lesions of the central nervous system (WHO grade IV), most frequently found intracranially in infants and young children.[8,5] ATRT was first documented in an adult who presented with a brain tumor in 1992; since then, it has rarely been reported intracranially or within the spinal canal.[6,2]

Establishing the pathological diagnosis of ATRT

The pathological diagnosis of ATRT entails the identification of sheet of rhabdoid cells and inactivation of the INI1 (SMARCB1) gene.[3] In addition, EMA, vimentin, and smooth muscle actin are typically diffusely positive.[4]

Imaging of ATRT

Imaging characteristics have been shown to be highly inconsistent including variable enhancement and cystic patterns.[7] They are found in the intradural space and do not typically invade extradurally nor exhibit bony invasion.

Three prior spinal cases of ATRT in adults

To our knowledge, there are only three cases of primary ATRT that involve the adult spine.[1,6,8] Bruch *et al*. was the first to report ATRT in a 21-year-old female; however, this was only a minimal description of the specific pathological specimen.[1] Secondarily, a 43-year-old female with confirmed ATRT involving both the cervical and lumbar canal was described; this report afforded a detailed description of the molecular, immunohistochemical, and cytogenetic characteristics.[8] Third, and more recently, a 65-year-old male presented with a primary ATRT involving the lumbar spinal canal, resulting in acute cauda equine syndrome.[6] Notably, the mean post diagnosis survival for these three patients was just 20 months.

CONCLUSION

ATRT only very rarely involves the adult spine, and must be differentiated from primitive neuroectodermal tumor (PNET). Owing to the aggressive nature of this disease, further reporting of this disease process is necessary for early tumor resection and adequate utilization of adjunctive treatment.

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Conflicts of interest

There are no conflicts of interest.

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