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

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].

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 later.

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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 2: Postoperative MRI. Clockwise from top left: T1 sagittal with 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

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
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 postoperative
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 actin. 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. 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.

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. In addition, EMA,
vimentin, and smooth muscle actin are typically diffusely
positive.

Imaging of ATRT
Imaging characteristics have been shown to be highly
inconsistent including variable enhancement and cystic
patterns. 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. 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. 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.
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. 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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