Brain & Development 39 (2017) 717–721

BRAIN & DEVELOPMENT
Official Journal of the Japanese Society of Child Neurology

www.elsevier.com/locate/braindev

Case Report

Extrarenal rhabdoid tumor presented with an immobile arm in a one-year-old boy

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Received 17 December 2016; received in revised form 30 March 2017; accepted 3 April 2017

Abstract

Infants with an immobile arm may be easily overlooked in primary care settings. Differential diagnoses include injuries, infections, neuropathies, ischemia and rarely, neoplasms. We report the case of a one-year-old boy with weakness in his left arm after minor trauma with a diagnosis of brachial plexus palsy initially. After rehabilitation for 2 months, his weakness progressed to unsteady gait and quadriparesis. MRI revealed a huge solid tumor in the left supraclavicular fossa, which also involved the left brachial plexus, upper thoracic cavity, and left paravertebral space with invasion into the spinal canal. Microscopically, the medium–large polygonal tumor cells had an eccentric eosinophilic cytoplasm and immunostaining showed a loss of nuclear INI1 expression. Array comparative genomic hybridization of the tumor tissue confirmed a segmental deletion at chromosome region 22q11.23 involving the *SMARCB1* gene. The final diagnosis was cervical paravertebral malignant rhabdoid tumor with intraspinal epidural and intradural invasion, a rare case of extrarenal extracranial rhabdoid tumor (ERRT). The intraspinal part of the tumor was resected followed by interval-compressed chemotherapy with vincristine–doxorubicin–cyclophosphamide alternating with ifosfamide–etoposide (VDC/IE). The tumor showed very good partial response to four cycles of chemotherapy with gradual recovery of neurological symptoms. ERRT is a very rare and aggressive tumor that mainly occurs in infants and children and may manifest with vague neurological symptoms when it involves the spinal cord and/or peripheral nerves. A neoplasm such as ERRT originating from or involving the brachial plexus should be considered in the differential diagnosis of an immobile arm in infancy.

Keywords: Immobile arm; Malignant rhabdoid tumors; Extrarenal extracranial rhabdoid tumor; Brachial plexus palsy; *SMARCB1* gene; INI1; Infant

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http://dx.doi.org/10.1016/j.braindev.2017.04.001
0387-7604/

1. Introduction

An immobile arm is relatively common in pediatric patients evaluated in both emergency departments and primary care clinics [1]. Because many young children are often afraid of strangers and cannot express themselves clearly, evaluating them can be challenging. Infants with weakness in one arm are often diagnosed as brachial plexus palsy and easily overlooked in rehabilitation and neurology clinics.

Malignant rhabdoid tumor (MRT) is a rare and highly aggressive tumor occurring in infancy and early childhood; it was first described in the kidneys in 1978 as a variant of Wilms tumor with rhabdomyosarcomatous characteristics [2]. It is currently called MRT in the kidneys, atypical teratoid/rhabdoid tumor (AT/RT) of the brain, and extrarenal, extracranial rhabdoid tumor (ERRT) in other sites of body such as soft tissue or liver [2,3]. ERRT is extremely rare and highly aggressive [4]; it predominantly involves a deep axial location, such as the neck or paraspinal region. We report a one-year-old boy developing ERRT of the lower neck, involving the brachial plexus, invading the spinal cord and paraspinal area.

2. Case report

A 14-month-old previously healthy boy was referred to our hospital with mild fever for 3 days and progressive left arm weakness for more than 2 months. His parents reported that he moved his left upper arm increasingly less frequently after a questionable minor trauma and viral infection 2–3 months ago. The weakness was mainly over the left shoulder and elbow joints, involving the proximal limb more prominently than the distal limb. Electromyography and nerve conduction velocity revealed a denervation pattern with fibrillation waves. A rehabilitation program was initiated immediately, and the parents felt that the patient's weakness improved thereafter.

Two months later, the patient's left upper limb weakness progressed to quadriparesis with an unsteady gait. Polyneuritis, Guillain–Barré syndrome, or tuberculosis was suspected. Cerebrospinal fluid (CSF) was evaluated, which showed elevated protein levels (300 g/dL) and pleocytosis (white blood cells, 95 per high-power field). All cultures, viral studies, and a tuberculosis test in the CSF and blood yielded negative results.

Chest X-ray revealed increased radiopacity superimposed on the left upper lung zone and scapular region (Fig. 1). A left axillary mass with widening of spinal canal from the cervical to upper thoracic spine was depicted by computed tomography (CT). Magnetic resonance imaging (MRI) of the cervical spine and neck revealed a $4 \times 3 \times 6$ cm multilobulated solid tumor occupying the left supraclavicular fossa. The tumor also involved the left brachial plexus, upper thoracic cavity, and the left paravertebral space with invasion into the cervical and upper thoracic spinal canal. The intraspinal component of the tumor expanded from C4 to T1 levels and resulted in remarkable compression and displacement of the adjacent spinal cord (Fig. 2A–C). Multiple CSF drop metastases were also observed in the lumbosacral region. Based on his clinical presentation and imaging characteristics, metastatic neuroblastoma or malignant neurofibrosarcoma should be suspected. However, the 24-h urinary vanillylmandelic acid and catecholamine levels were within reference ranges.

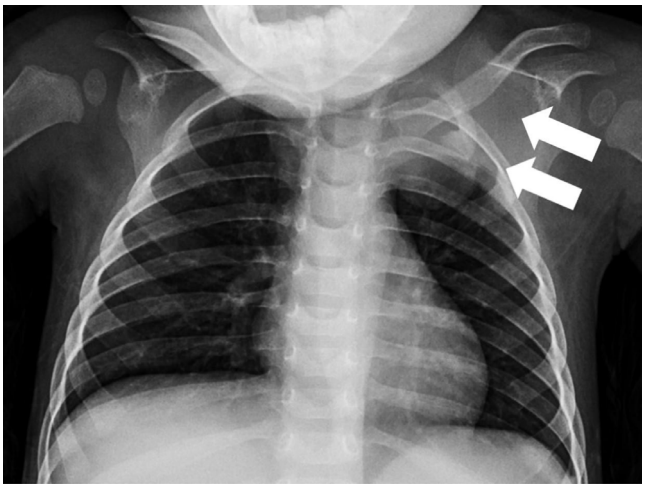

Fig. 1. Chest X-ray performed at diagnosis showed focally increased radiopacity superimposed on the left upper lung zone and scapula (arrows).

An emergent neurosurgical procedure, namely C4–C7 laminotomy with resection of the epidural and intradural extramedullary tumor was performed under intraoperative neurophysiological monitoring. The epidural and intradural parts of the tumor extending from the C5 root to the C7 root were resected, whereas the lateral part of the tumor involving the intervertebral foramina and left brachial plexus was retained. Histologically, the tumor was composed of medium–large polygonal tumor cells with vesicular and prominent nucleoli in sheet-like patterns; some of the cells had an eccentric eosinophilic cytoplasm (rhabdoid cells). Immunohistochemically, the tumor was positive for vimentin (diffuse), cytokeratin, CD99, and S-100 but negative for Melan-A, myogenin, glial fibrillary acidic protein, synaptophysin, common leukocyte antigen, and human melanoma black 45. The integrase interactor 1 (INI1; encoded by *SMARCB1*) protein expression was negative, and the Ki-67 index was 10%. Array comparative genomic hybridization of the tumor tissue confirmed a segmental deletion at chromosome region 22q11.23 involving the *SMARCB1* gene (Fig. 3A–F). Altogether, an ERRT was the diagnosis.