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Case report

A case of intraneural perineurioma presenting with monomelic atrophy in a child

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Abstract

We report the case of an 11-year-old girl who developed slowly progressive atrophy of the left lower extremity. She suffered from mild dilated cardiomyopathy of unknown cause since 4 years of age. When she was 7 years old, her family noticed that her left extremity was thinner compared to the right one. Computed tomography showed atrophy and areas of low density in the left gluteus maximus, thigh, and calf muscles. The left sciatic nerve showed gadolinium enhancement on magnetic resonance imaging. A biopsy of the left sural nerve revealed pseudo-onion bulbs. Immunohistochemical staining was positive for epithelial membrane antigen and negative for S100 protein. Electron microscopy demonstrated myelinated or unmyelinated nerve fibers surrounded by concentric layers of perineurial cells. These results indicated intraneural perineurioma. The tumor was estimated at least from the nerve root to the ankle joint. The length of nerve involvement in this patient was the highest recorded in the literatures. Intraneural perineurioma is a very rare disorder, but is tend to be found in youth. This disorder should be considered when we see children with monomelic weakness and/or atrophy.

Keywords: Intraneural perineurioma; Monomelic atrophy; Benign tumor; Peripheral nerve

1. Introduction

Although monomelic atrophy is rare, many underlying disorders are known [1]. Hirayama disease affecting the upper extremity [2] is well known. In the lower extremities, the etiology in most patients with monomelic atrophy is neurogenic, e.g., tethered cord, liability to pressure, entrapment syndromes, etc. [1]. Muscular disorders originating in monomelic atrophy are rarely reported [3,4]. Recently, monomelic atrophy caused by benign tumors in peripheral nerves has been reported [1]. We report a case of a very long intraneuronal perineurioma in a child, whose left lower extremity showed slowly progressive atrophy for 4 years.

2. Case report

The patient was an 11-year-old girl. Her parents were healthy, her delivery was uneventful, and her early development was normal. She suffered from mild dilated cardiomyopathy since 4 years of age. The etiology was unknown and medical treatment was not needed. At

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7 years of age, her parents noticed a left foot drop when she walked. There was no history of trauma or antecedent illness. She went to see an orthopedic specialist, who pointed out the shortness of her left lower extremity and pes equinovarus in her left foot. At 9 years of age, her foot deformity had progressed and she had to raise her left knee while walking. Lumbosacral plexus neuropathy was suspected. She underwent intravenous γ-globulin therapy, which resulted in no improvement. At 11 years of age, she was admitted to our hospital.

She was well nourished and moderately structured (149 cm, 37 kg). The left lower extremity was shorter and thinner than the right one. The length of the left lower extremity was 68 cm and that of the right one was 72 cm. Mid-thigh circumference was 36 cm on the left and 39 cm on the right side. The circumferences of the left and right calves were 24 cm and 28 cm, respectively. There was no discrepancy between the upper extremities. Physical examination was normal except for the contracture in the left ankle joint, which could not be dorsiflexed. Heart rate was not accelerated. Neurologic examination showed absence of the left achilles tendon reflex, probably because of the contracture, and a distal-dominant painless muscle weakness of the left lower extremity. Sensory disturbance was not detected. The remainder of the neurologic examination was normal.

Blood chemistry, including creatine kinase, lactic acid, and pyruvic acid, was normal. A nerve conduction velocity study was performed. Motor conduction velocity (MCV) and sensory conduction velocity (SCV) of the median and tibial nerves were within normal limits on both sides. SCV in the right sural nerve was normal. SCV in the left sural nerve was not detected.

Computed tomography (CT) of the muscles revealed a decreased volume of the left gluteus, thigh, and calf muscles (Fig. 1). Magnetic resonance imaging (MRI) showed gadolinium enhancement of the hypertrophied left sciatic nerve from the nerve root (Fig. 2).

2.1. Histology and immunohistochemistry

Sural nerve biopsy was performed at the left ankle. Hematoxylin–eosin-stained sections showed a plexiform pattern of expanded nerve fascicles embedded within a fibrovascular stroma (Fig. 3a). The expansion consisted of a proliferation of pseudo-onion bulbs with interspersed collagen bundles. Immunohistologic staining was positive for epithelial membrane antigen (EMA) (Fig. 3b) and negative for S100 protein (Schwann cell marker) (Figs. 3c), indicating that the spindle cells were of perineurial origin.

Electron microscopy revealed that the pseudo-onion bulbs consisted of myelinated or unmyelinated nerve fibers surrounded by concentric layers of perineurial cells (Fig. 3d).

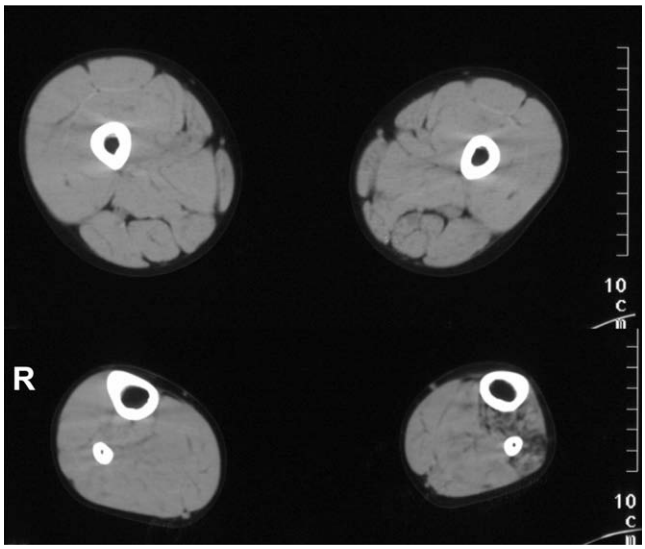

Fig. 1. Muscle CT. The volumes of the left thigh (upper) and calf muscle (lower) are less than those of the corresponding muscles on the right.

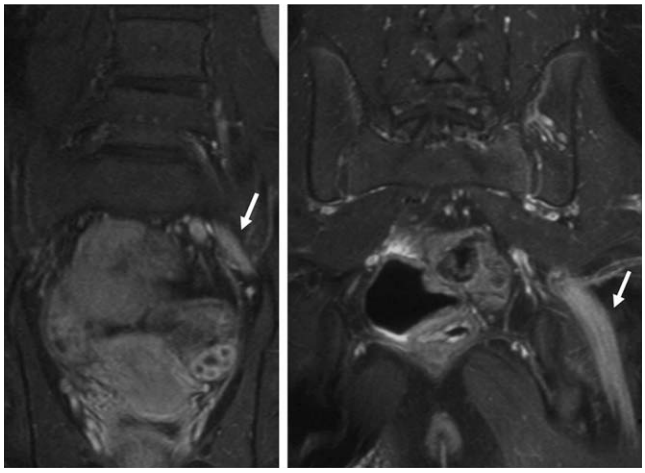

Fig. 2. MRI. Gadolinium-enhanced T1-weighted images. The left lumbosacral enlarged nerve root is enhanced by gadolinium (white arrows).

The patient was diagnosed as having intraneural perineurioma. The tumor was estimated to extend at least from the nerve roots in the left lumbosacral plexus to the left sural nerve at the ankle level. Because of the great length of this intraneural perineurioma, no adequate treatment could be provided.

3. Discussion

Intraneural perineurioma is a rare and benign type of peripheral nerve tumor. The relationship between intraneural perineurioma and dilated cardiomyopathy is not known. Perineurioma is histologically subclassified as intraneural and extraneural [5,6]. If the perineurial cell proliferation is confined within a nerve, it is named