Neuromuscular Choristoma of the Sciatic Nerve
- A Case Report -

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Neuromuscular choristoma is a rare benign tumor of the peripheral nerves. To the best of our knowledge, 21 cases have been reported to date. We describe here a 20-day-old female infant who presented with a buttock mass (4.5 × 4.1 × 3.2 cm on MRI) arising from the left sciatic nerve. Microscopically, it was characterized by an intimately disorganized mixture of nerve fibers and striated muscle fibers that were occasionally surrounded by the perineurium and separated by fibrous bands of varying thickness. In some areas, there appeared to be some cells in transitional forms between nerve fibers and muscle fibers, revealing variously positive expressions for S-100 protein in the muscular components. These findings are consistent with the hypothesis that neuroectodermal-derived Schwann cells can give rise to mature skeletal muscle. It appears that the fibrosis may be related to the degeneration of the neural components. The size of the mass on MRI has been unchanged during the 3-year follow-up period.

Key Words : Neuromuscular choristoma; Sciatic nerve; Striated muscle fiber; Schwann cell

CASE REPORT

A 20-day-old female infant presented with a left buttock mass that had been noted since birth. The infant’s physical examination was normal. There was no pertinent family history. The MRI revealed a well-circumscribed mass in the left buttock, 45 × 41 × 32 mm in size. The axial T1- (Fig. 1A) and T2-weighted images (Fig. 1B) showed lower signal intensity for the tumor than that of the muscle. After a contrast infusion, there was mild heterogeneous enhancement. The left sciatic nerve was not clearly defined as compared with the right side (Fig. 1). During surgery, the elements of the left sciatic nerve appeared to enter into the center of the buttock mass. Biopsy was done, but excision was not performed. Over a 3-year period following biopsy, no significant change in the size of the lesion has been noted on MRI.

The biopsy specimens were fixed using formalin and then embedded in paraffin. Sections were stained with hematoxylin and eosin, Masson’s trichrome, and phosphotungstic acid-hematoxylin (PTAH), and the sections were also treated for immunohistochemical markers of S-100 protein, desmin, and epithelial membrane antigen (EMA). Microscopically, the tumor showed small nodules and fascicles subdivided by dense fibrous tissue, which were composed of well-developed striated muscle fibers and myelinated and nonmyelinated nerve fibers in various pro-
portions (Fig. 2A, B). The nodules were encompassed by the overlapped layers of EMA-positive flattened perineurial cells (Fig. 2C). The two basic components of nerve fibers and muscle fibers were indivisibly intermingled, and these two types of fibers were confirmed by immunostaining for S-100 protein and desmin, respectively (Fig. 2D, E). There were occasional areas that seemed to display the transition of nerve fibers to striated muscle fibers (Fig. 3A). The immunostaining for S-100 protein revealed various levels of positivity in the muscular components within the nodules (Fig. 3B). The muscle fibers showed round, ovoid and spindle configurations, and the cytoplasm revealed cross-striations. Muscular differentiation or maturation became more evident at the outside of the nodules. In some areas, the nerve fibers and muscle fibers were individually scattered in a collagenous stroma. These randomly scattered cells appeared to originate from the fascicles or from the break-up of the nodules. Dense

Fig. 1. Axial T1W1 (A) and T2W1 (B) of the left buttock show a well-circumscribed low signal intensity mass under the gluteus muscle in the left buttock (arrows). The left sciatic nerve is not clearly defined as compared with that of the right side (arrow head).

Fig. 2. (A) Neuromuscular choristoma shows multinodular character of the lesion surrounded by various fibrosis (Masson trichrome). (B) The typical nodule is composed of nerve fibers and striated muscle fibers. (C) The nodule is surrounded by the overlapped layer of EMA-positive perineurium (arrows). (D) Immunostaining for S-100 shows positive nerve fibers within the nodule. (E) Immunostaining for desmin shows positive muscle fibers within the nodule.
Fibrous tissue was admixed in various degrees and it replaced the muscular and neural components (Fig. 1). The muscular components showed atrophy, apoptosis or degenerative change without any regenerative fibers (Fig. 4), and the small nerve bundles showed focally myxoid degeneration. Neither a rhabdomyoma nor any suggestion of malignancy has been observed in the tissue samples.

**DISCUSSION**

Neuromuscular choristomas or hamartomas are benign tumors that are composed of skeletal muscle and neural elements, and these tumors generally occur in infants and young children. Triton tumor refers to a peripheral nerve sheath tumor in which the muscle differentiates from the neoplastic nerve sheath cells that are, presumably, neoplastic Schwann cells. However, benign triton tumor is often loosely used to refer to the neuromuscular choristomas in accord with an early hypothesis concerning their histogenesis. To date, 21 cases of neuromuscular choristoma have been reported in the world medical literature. These tumors usually arise in association with large nerves such as the brachial plexus, the sciatic nerve, and the proximal median nerve, although several examples involving the spinal intradural space and the skin have been described. Depending on their sizes, locations and degrees of impingement on surrounding normal structures, the patients with this tumor have experienced minimal to striking disabilities. Our case is the sixth case involving the sciatic nerve, and the clinical backgrounds of the other five patients are summarized in Table 1. Of the five patients described in the literature, four had foot deformities. These findings emphasize the need for the careful evaluation of those patients who show a unilateral deformity of the foot.

The histogenesis of neuromuscular choristoma remains a controversial issue. Louhimo and Rapola suggested that the skeletal muscle might originate from the limb mesenchymal cells trapped within the nerve sheath during embryogenesis. Another possibility is that the neuroectoderm, the so-called ectomesenchyme, can give rise to mature skeletal muscle in much the same way as the neuroectodermal eye cup gives rise to the iris muscle. Masson has offered that endoneurial cells of neuromas, under the organizing influence of the motor nerve fibers, may be able to differentiate into muscular tissue and this hypothesis was used to explain the histogenesis of malignant triton tumor. Karyotypic analysis in one of the previous case was normal, and this favors the opinion that this lesion is a malformation and not a true neoplasm. The histological features of

![Fig. 3.](image1.png) **Fig. 3.** (A) High power view of the nodule shows intimately disorganized admixture of nerve fibers and striated muscle fibers. There are some areas seemed to be transitional forms between these two fibers. (B) Immunostaining for S-100 shows various positivity in the muscular component (arrows).

![Fig. 4.](image2.png) **Fig. 4.** There are atrophy, apoptosis or degenerative changes of muscle fibers.

![Fig. 5.](image3.png) **Fig. 5.** There are atrophy, apoptosis or degenerative changes of muscle fibers.
our case were similar to those of the previously reported cases of neuromuscular choristomas (hamartomas). The lesion of our case showed both nerve fibers and striated muscle fibers in various proportions and the two basic components were indivisibly intermingled in a way that was probably related to their common histogenesis. A variable number of Schwannian cells were seen to merge with striated muscle cells, and the immunostaining for S-100 protein revealed variously positive expressions in the muscular components. These findings support the hypothesis that the development of striated muscle is the metaplastic versatility of the Schwann cell or of its progenitor, the neural crest cell.

At times, the fibrous component surrounding the lesions was very dense and cellular, which suggests the diagnosis of fibromatosis replacing muscle and nerve. In three previous instances (two nondiagnostic biopsies and one gross total resection), the procedures were followed by postoperative fibromatosis. It appears that the fibrosis may be related to degeneration of the neural components. The factors that influence the mesenchymal differentiation into skeletal muscles are not completely understood, but in general, the initial differentiation of muscle is independent of neural influences. However, environmental and neural influences are important factors that play a role in determining the extent of muscle growth, organization, and maintenance.

In neuromuscular choristoma, the nodules encompassed by a perineurium can be expanded by the muscular proliferation or replacement, and when the perineurium is broken-up, they form fascicles. The neural components may also degenerate or be lost. When the trophic stimuli to muscle differentiation stops, the muscular components then degenerate or become atrophic. Both the degenerated neural and muscular components are replaced by fibrosis. This process may be related to spontaneous regression or to the postoperative fibromatosis together with the inciting factors, which have been most commonly associated with prior surgery and irradiation. Our case as in other cases revealed that the dense fibrous tissue mixed to varying degrees and replaced the muscular and neural components.

In some areas, the nerve fibers and muscle fibers were individually scattered in a collageinous stroma. These randomly distributed cells appear to have originated from the fascicles or from the break-up of nodules encompassed by overlapped EMA-positive perineurial cells. The degenerative changes of the muscular components and the small nerve bundles noted in this case suggest that the fibrosis may be initially induced by the degeneration of the neural components.

The follow-up information in our case and of those cases in the literature supports the benignity of this tumor. The cases involving the sciatic nerve showed regression, fibromatosis or a persistent neurologic deficit (Table 1). Although resection is curative, biopsy or incomplete excision has resulted in the amelioration of symptoms and progressive decrease in tumor size. Therefore, after a correct diagnosis is achieved, the proper

<table>
<thead>
<tr>
<th>Authors</th>
<th>Sex/Age (yr)</th>
<th>Location and size (cm)</th>
<th>Symptom (neurologic deficit)</th>
<th>Treatment</th>
<th>Follow-up</th>
</tr>
</thead>
<tbody>
<tr>
<td>Louhimo and Rapola (1972)</td>
<td>M/birth</td>
<td>L: 4</td>
<td>No</td>
<td>PR at 4 mo old</td>
<td>Regression of residual lesion</td>
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<td>Markel and Enzinger (1982)</td>
<td>M/15 mo</td>
<td>R (buttock)</td>
<td>Valgus and overextended toes</td>
<td>PR at 2 yr old</td>
<td>No follow-up</td>
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<td></td>
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<td>Gastrocnemius and posterior</td>
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<td></td>
<td></td>
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<td>Tibial muscle paralysis</td>
<td></td>
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<tr>
<td>Boman et al. (1991)</td>
<td>F/4</td>
<td>R</td>
<td>R foot equinus</td>
<td>B (nondiagnostic)</td>
<td>Postoperative fibromatosis</td>
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<td></td>
<td>5</td>
<td>R</td>
<td>R leg spasticity</td>
<td>PR of fibromatosis</td>
<td>Regrowth of fibromatosis with deterioration in R leg function</td>
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<td></td>
<td>7</td>
<td>R</td>
<td></td>
<td>Amputation (fibromatosis with accompanying choristoma)</td>
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<tr>
<td>Bassett et al. (1997)</td>
<td>M/8</td>
<td>R (the pelvis to the ischia tuberosity): 3 × 8 × 2.5</td>
<td>R foot cavovarus deformity</td>
<td>B</td>
<td>Persistent neurological state</td>
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<td>Atrophy and weakness of</td>
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<td></td>
<td></td>
<td></td>
<td>R lower extremity</td>
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<tr>
<td>Maher et al. (2002)</td>
<td>M/18</td>
<td>R (from the 2nd sacral nerve root down the thigh): 23 × 3</td>
<td>Atrophy and weakness of</td>
<td>B</td>
<td>Persistent neurological state</td>
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<td>R lower limb</td>
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<td></td>
<td>Talipes cavus and hammertoe deformities of R foot</td>
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<td></td>
</tr>
<tr>
<td>Present case</td>
<td>F/20 day</td>
<td>L (buttock) : 4</td>
<td>No</td>
<td>B</td>
<td>3 yr: no change in the size</td>
</tr>
</tbody>
</table>

R, right; L, left; mo, month; yr, year; PR, partial resection; B, biopsy.
treatment should be conservative and aimed primarily at maintaining the integrity of the nerves.  

REFERENCES