CASE REPORT

Cutaneous neuromuscular choristoma: Case report and review of the literature

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A B S T R A C T

Neuromuscular choristoma, also known as benign triton tumor, is an uncommon developmental tumor composed of skeletal muscle and neural elements. The majority of cases reported have occurred in infants and have been generally associated with a large nerve, such as the brachial plexus or sciatic nerve. To date, only three cases of cutaneous lesions have been reported in the English literature. We describe a 28-year-old female presented with a soft, pedunculated papule on the presternal region since birth and review the literature of neuromuscular choristoma.

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Introduction

Neuromuscular choristoma, also known as benign triton tumor, is composed of skeletal muscle and neural elements. It is an uncommon benign tumor. The majority of cases reported have occurred in infants and have been generally associated with a large nerve, such as the brachial plexus1–9 or sciatic nerve.1,2,10–13 Cranial nerve involvement has been reported in subsequent English-language literature.14–30 Cutaneous lesions are very rare.31–33 Here, we describe a new case that presented with soft, pedunculated polypoid skin on the presternal region since birth.

Case report

A 28-year-old healthy woman presented with polypoid skin on the presternal region that had been present since birth (Figure 1). The lesion was asymptomatic and measured 6 mm × 6 mm × 4 mm. The lesion was not growing and there was no neurological deficit. Shave excision was performed under the impression of a skin tag or a branchial arch remnant.

The biopsy specimen was fixed with formalin and then embedded in paraffin. Sections were stained with hematoxylin and eosin and an immunohistochemical study was performed with antibodies including S-100 protein, desmin and HHF-35. Microscopically, it was a polypoid skin tumor with bundles of perpendicularly oriented skeletal muscles, intermingled with multiple small nerve fascicles in the center and surrounded by groups of eccrine sweat glands in the periphery (Figure 2A and 2B). Blood vessels were also present. No nuclear atypia, mitoses or major nerve bundles were observed. The skeletal muscle fibers were strongly positive for desmin (Figure 2C) and HHF-35. Nerves were positively stained by S-100 protein (Figure 2D). Skeletal muscles were also stained by S-100 protein, to a lesser intensity than nerves. The histopathological picture was characteristic of a neuromuscular choristoma or benign triton tumor and the immunohistochemical staining confirmed the presence of skeletal muscles and nerves.

Discussion

Neuromuscular choristoma is a benign tumor composed of skeletal muscle and neural elements. Louhimo and Rapola1 first reported two cases in 1972 and they regarded these tumors as intraneural muscular hamartoma. However, hamartomas are defined as benign tumor-like nodules composed of an overgrowth of mature cells and tissues, but with disorganization and often with one element predominating. Choristoma is defined as a mass formed by the faulty development of tissue of a certain type not normally found at that site. In 1983, Bonneau and Brochu3 used the term “neuromuscular choristoma” because the muscle fibers could not be considered as normal constituents of peripheral nerves. It is a rare benign tumor that most often occurs in the first decade of life. To date, only 39 cases have been reported in the English-language literature. The total number of cases was underestimated in previous review articles because of the different nomenclature. The clinical
The clinical backgrounds of reported cutaneous neuromuscular choristoma are summarized in Table 1. Our patient is a new case without major nerve involvement.

O’Connell and Rosenberg first described two cutaneous lesions from a 3-month-old female infant in 1990. These lesions, each measuring 1 cm × 1 cm × 0.9 cm, were polypoid and located in the submandibular skin and over the sternum, respectively. Both lesions were removed surgically, and neither was associated with a major nerve. The second case was identified in the lumbosacral lipoma of a 4-month-old boy. Demir et al. reported a 17-year-old girl with a solid, nontender, immobile 2.5 cm × 2 cm mass with irregular borders located at the left mental region, present since early childhood. Slight weakness at depression of the lower lip in the affected side was noted during motor examination. A mixture of well-differentiated skeletal muscle cells and nerve fibers was demonstrated in these three cases. If there is fat tissue in the lesion, the possibility of rhabdomyomatous mesenchymal hamartoma should be also considered. With the exception of the one arising from lipoma, there was neither fat in the lesion, nor adjacent dermis, nor major nerve. In contrast, striated muscle
The age range is wide, from newborn to 74 years.

A choristoma should be considered as an isolated entity. There have been only two cases accompanied with fibromatosis, and another two cases associated with lymphangioma and Freeman–Sheldon syndrome. The age range is wide, from newborn to 74 years. Neuromuscular choristomas with peripheral nerve involvement (13/16) and cutaneous involvement (4/4) were more common in children. The majority of the cases (55%) were detected before the age of 1 year and 80% of cases were found before the age of 5 years. However, 12 of the 18 cases (66%) of intracranial neuromuscular choristoma occurred in teenagers and adults. Despite the absence of obvious sexual predilection, the male–female ratio was 1:3 in cutaneous neuromuscular choristoma.

The histogenesis of neuromuscular choristoma remains controversial. Several theories have been suggested to explain the mixture of well-differentiated skeletal muscle cells and nerve fibers in this tumor and the probable common histogenesis. Louhimo and Rapola have suggested that undifferentiated mesenchymal tissue incorporates into neuromas as the embryonic nerves extend peripherally, and entrapped mesenchymal tissue might later develop into skeletal muscles. Markel and Enzinger have stated that mesenchymal tissue might be derived from neuroectodermal cells, which could give rise to mature skeletal muscle in the same way as the neuroectodermal eye cup gives rise to the iris muscle. Kusuzaki has suggested that the structure of the neuromuscular junction may be similar to that in the motor end-plate of the normal muscle, but it may not be functional, because the hamartomatous muscles cannot contract by nerve stimulation. Immunostaining for S-100 protein revealed positive expression in the muscular components, therefore, Kim has suggested that development of striated muscle is the metaplastic versatility of Schwann cells or of their progenitors, the neural crest cells. However, S-100 protein could stain many different cells, such as melanocytes and Schwann cells, eponymous cells, and smooth and skeletal muscle. It is a commonly used immunohistochemical stain for melanocytic and neural neoplasms because of its high sensitivity. Therefore, the positive expressions of S-100 protein in the muscular components should not imply Schwann cell or neural crest cell origin.

Sometimes, total excision of neuromuscular choristoma is impossible because of its close association with a major nerve and the possible sequelae of functional deficits, such as postoperative distal motor paralysis, facial palsy, and diabetes insipidus. Therefore, only partial excision or biopsy of the lesion can be performed in many cases. Spontaneous regression of the tumor after biopsy and partial resection have been reported in patients with peripheral nerve involvement. Chen has suggested that the only surgical procedure required is a biopsy to establish the diagnosis. Complete excision is more practical in cutaneous lesions because they have no relationship with major nerves. Local recurrence is uncommon because of its benign clinical behavior.

In addition, a branchial arch remnant was suspected clinically. Nevertheless, branchial arch remnants should present as subcutaneous cartilaginous tags along the anterior border of the sternocleidomastoid muscle. The preternal location of our patient did not favor a diagnosis of branchial arch remnant. Despite their low incidence, branchial arch remnants should be taken into account when evaluating skin-tag-like lesions on the neck.

In conclusion, neuromuscular choristoma is an uncommon benign tumor mainly associated with major nerves. The histogenesis is still unclear. Cutaneous neuromuscular choristoma is extremely rare. Here, we report the fourth cutaneous case and propose the early onset and female predominance. In contrast to the difficulties in resection of neuromuscular choristoma, cutaneous lesions can be managed more easily because they are free of major nerve involvement.

References


