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Long-term natural history of a neuromuscular choristoma of the sciatic nerve: a case report and literature review

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ABSTRACT

Neuromuscular choristomas are rare tumors with mature skeletal muscle elements admixed with neural elements. Follow-up results are limited, the natural history of neuromuscular choristomas is poorly understood. To date, 14 cases of sciatic nerve neuromuscular choristomas have been reported, but follow-up results were provided only for 10 cases (median duration: 2 years). The natural history of neuromuscular choristomas is therefore poorly understood due to lack of longitudinal data. We describe a case with long-term natural history for 17 years of the disease and provide a literature review of the reported cases.

A 6 year old girl, with a 6 year history of left buttock mass with neonatal pathologic diagnosis of neuromusculoar choristoma, presented with intermittent left lower extremity weakness in July 2006. She had a left buttock mass for 6 years, and the neonatal pathologic diagnosis was neuromuscular choristoma. Magnetic resonance imaging (MRI) revealed fusiform enlargement of the left sciatic nerve with mild enhancement similar to that of the muscle. The pathology of a re-biopsy sample was also consistent with neuromuscular.

choristoma. In follow up, the mass increased slightly, but MRI findings remained consistent, and her condition remained stable for 11 years after re-biopsy. Aggressive fibrosis known as complication by manipulation of neuromuscular choristoma was also not developed. The patient has been followed up for a total of 17 years and this is the only case of long-term natural history of neuromuscular choristoma.

1. Introduction

Neuromuscular choristomas (NMCs), also known as benign triton tumors or neuromuscular hamartomas, are rare tumors in which well-differentiated muscle fibers are mixed with mature nerve fibers [1,2]. They typically involve the cranial or large peripheral nerves, such as the sciatic nerve or brachial plexus [3,4]. NMCs are known to be benign and slowly progressive, but given the lack of follow-up information, their natural history is poorly understood [5]. To date, 14 cases of NMCs of the sciatic nerve have been reported [1–4,6–12]. None of the reported cases included a long-term natural history of NMC. Herein, we report a case of NMC of the sciatic nerve and its long-term natural history for 17 years. We also present a review of 15 cases including the present case.

2. Case report

2.1. History

In July 2006, a 6-year-old girl presented with intermittent left lower extremity weakness. The patient had a 6.4-cm-sized left buttock mass for 6 years and complained about tenderness on palpation of the left buttock area without radiation of pain. The mass was hard and firm on palpation. The patient had mild muscle atrophy throughout the left buttock and posterior thigh. There was no limb length discrepancy. She did not exhibit skin lesions overlying the left buttock or elsewhere on her body, and no evidence of neurofibromatosis was noted. The lesion was discovered at birth and was 4.5 cm in size. Biopsy was performed 20 days after birth at another institution, and the mass was diagnosed as an NMC. Thereafter, the patient did not undergo any treatment or follow-up imaging study until July 2006.

2.2. Radiologic findings and pathology

In 2006, pelvic plain radiography demonstrated a soft tissue mass involving the left buttock and thigh, but the underlying bone appeared uninvolved (Fig. 1). Magnetic resonance imaging (MRI) image (Fig. 2) revealed a well-circumscribed, fusiform enlargement of the left sciatic nerve that extended 6.4 cm from the sciatic notch to the left thigh. The maximum transverse diameter of the sciatic nerve was 5.9 cm in the

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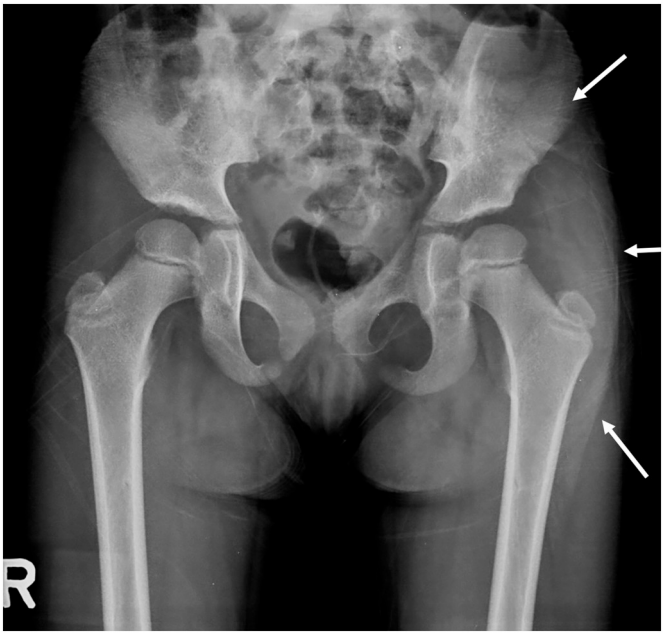

Fig. 1. A 6-year-old girl with neuromuscular choristoma in the left sciatic nerve. A pelvic radiograph demonstrates mild increased density bulging contour mass in the left buttock and thigh (arrows) without mineralization. The underlying bone appears uninvolved.

region immediately posterior to the greater trochanter. The fusiform enlargement appeared as low-signal intensities on T1- and T2-weighted images and mildly heterogenous enhancement similar to that of the muscle, including a striated appearance along the axis of the sciatic nerve. Longitudinal fibrous bands of hypo-intense T1- and T2-weighted images were present within the lesion. Mild muscle atrophy was observed in the left piriformis and gluteus maximus.

The patient had previously undergone a biopsy 5 years prior. However, we decided to perform a re-biopsy because the size of the lesion increased and symptoms developed. The patient underwent a surgical biopsy through exposure of the left sciatic nerve via a gluteus maximus-splitting approach. The proximal portion of the sciatic nerve appeared to be involved. The size of the mass was about 7 cm, well capsulated, with enlarged fusiform, hypertrophic, and firm. Neurolysis showed no evidence of non-muscular and non-neuronal tissue among the fibrous fascicles. The operating surgeon attempted aspiration using a syringe in the mass, but this was unsuccessful. Some fascicles were selected for biopsy. Postoperatively, she was neurologically stable and showed improvement of symptoms. The tissue specimen was embedded in paraffin and stained with hematoxylin and eosin. The specimen showed mature skeletal muscle fibers and enlarged nerve fascicles surrounded by dense fibrous tissue. The pathology was also revealed to resemble that of NMCs (Fig. 3).

2.3. Long-term follow-up

At the 7-month post-biopsy follow-up evaluation, an ultrasound image demonstrated no change in the size of the mass (Fig. 4). She underwent a total of six ultrasound examinations every 6 months until January 2010. Subsequently, the patient was examined once a year. An ultrasound examination in 2013 showed that the size of the mass slightly increased to 7.46 cm and there was little change thereafter. In July 2016, follow-up MRI was performed, which showed an increase in size to 8.8 cm (transverse diameter: 7.3 cm). Compared with those shown in the previous MRI in 2006, the fusiform shape and signal intensities were the same. After 18 months, one more follow-up MRI in January 2018 (Fig. 5) was performed. The size (length: 9.0 cm; transverse diameter: 7.5 cm), shape, and signal intensities of the lesion were similar compared with those of lesion observed during the penultimate MRI.

Recently, the association between NMCs and aggressive fibromatosis has been reported. Biopsy or surgical manipulation are considered to lead to the occurrence of aggressive fibromatosis [1,8]. Our patient had no images in which findings of aggressive fibromatosis were suspected. The patient exhibited no new neurological deficits and was stable, both subjectively and objectively. She will be re-evaluated yearly with repeat MRI scans to monitor the progression of the buttock lesion and development of aggressive fibromatosis.

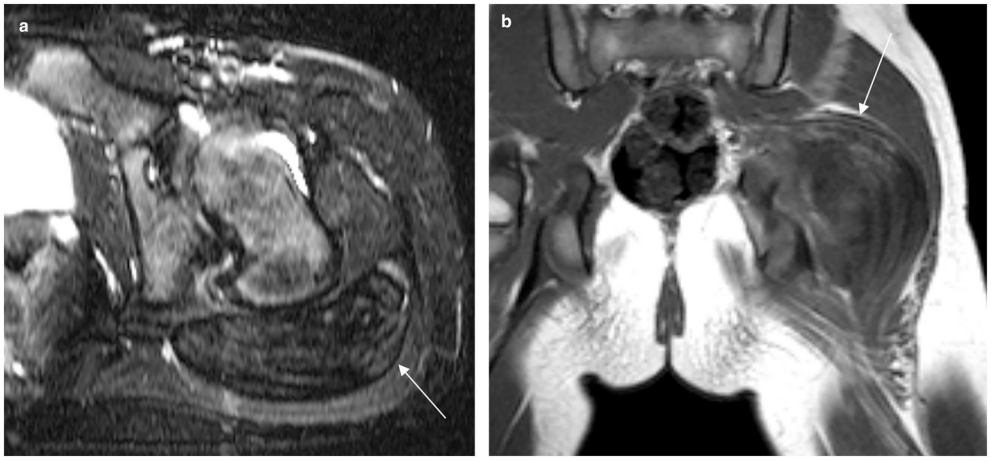

Fig. 2. a Axial T2-weighted turbo spin echo (TSE) magnetic resonance image (MRI) with fat suppression (FS) (repetition time (TR):4870 echo time (TE):99) shows the round mass to be hyper-intense in signal (arrow). b Coronal T1-weighted MRI (TR:4300 TE:14) shows well-circumscribed, fusiform enlargement of the left sciatic nerve (arrow) that extended from the sciatic notch to the left thigh.

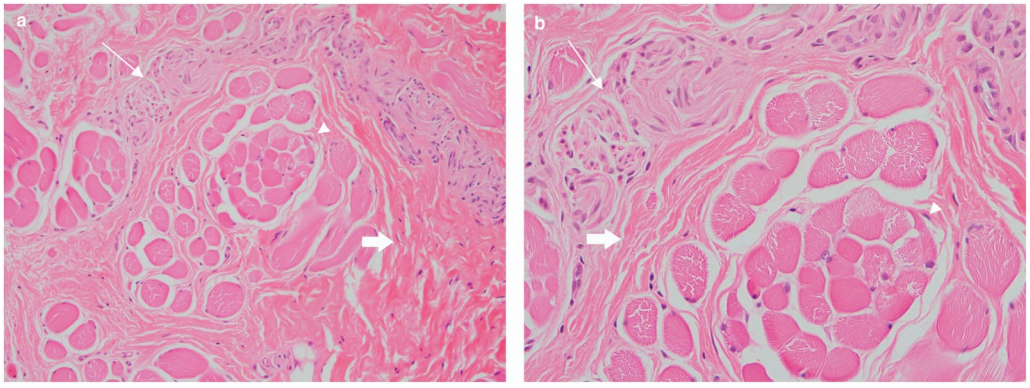

Fig. 3. Specimens obtained at biopsy: Mature skeletal muscle fibers (arrow heads) and enlarged nerve fascicles (arrows) surrounded by dense fibrous tissue (wide arrows). (Hematoxylin and eosin staining: a magnification ×200, b ×400).

3. Discussion

NMCs are rare lesions in which well-differentiated muscle is found within nerves, such as the cranial, brachial plexus, or sciatic nerves [8]. Orlandi [11] reported the first description of sciatic nerve NMC in 1895. To our knowledge, our case is the 15th case of sciatic nerve NMC (Table 1). Our patient had a mass in the buttock at birth and was diagnosed with NMC based on a biopsy at that time. She was then lost to follow-up. At 6 years of age, she complained of weakness and underwent re-biopsy due to enlargement of the mass. The pathology was also revealed a NMC. Since then, she has been followed up for 11 years after re-biopsy until now: a total of 17 years. The size of the mass has grown slightly during follow-up. However, the MRI findings including signal intensity, shape, and degree of enhancement remain the same as those observed before.

The age of diagnosis, symptoms, radiologic findings, interventions, and natural history of the reported sciatic nerve NMCs are described in this literature review (Table 1). Most cases of NMCs occur in childhood. Three adult patients aged 68, 42, and 45 years have been reported, and 12 cases involved pediatric patients (age range: 0–18 years). The reported cases included six female and nine male patients. There were five cases of left-sided NMCs, nine cases of right-sided NMCs, and one case for which the orientation was not reported. The symptoms that most patients complained of was weakness (as in our patient; eight cases); other presenting symptoms in the reported cases included pain (five cases), cavovarus foot (four cases), short leg of the affected limb (four cases), atrophy (three cases), limp (three cases), spasticity (one case), and a palpable mass at birth (one case). Ten patients, including our patient, initially underwent MRI, which revealed well-circumscribed, fusiform enlargement of the affected sciatic nerve. The lesions

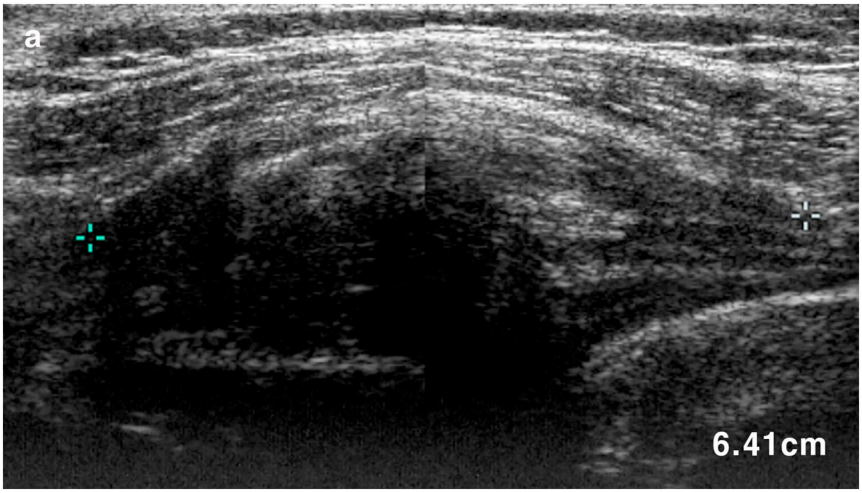

Fig. 4. At 7-months after re-biopsy, a follow-up ultrasound image demonstrates no change in the size of the mass (6.41 cm) compared with MRI in July 2006. A follow-up ultrasound in February 2013, the size of the mass slightly increased to 7.46 cm.

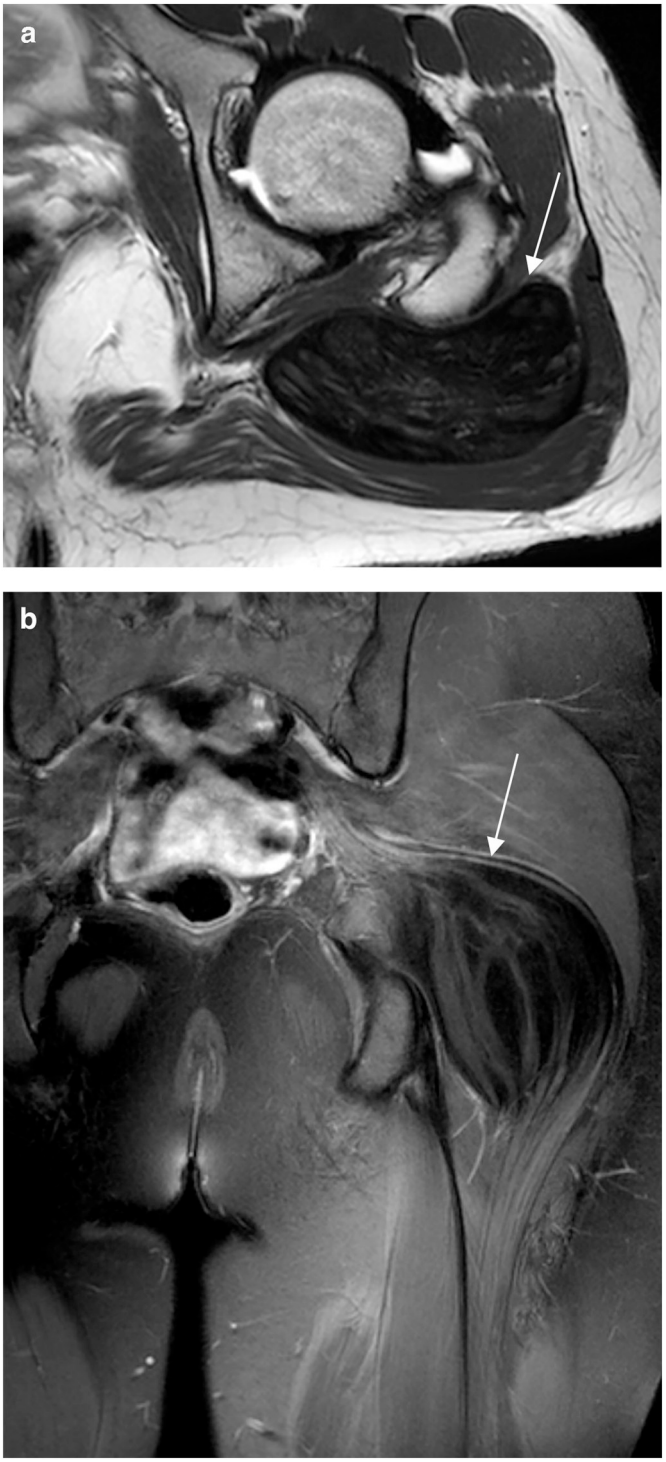

Fig. 5. Follow-up MRI images in January 2018. a Axial T2-weighed TSE (TR: 3740 TE: 80) shows round masses in the left buttock (transverse diameter: 7.5 cm). b Coronal T2-weighted spectral attenuated inversion recovery (SPAIR) with FS (TR: 2215 TE: 70) shows fusiform enlargement of the left sciatic nerve (length: 9.0 cm).

tended to appear with low signal intensity, similar to that of neighboring muscle, on T1- and T2-weighted images and mild heterogenous enhancement on post-contrast images. The first reported case was diagnosed at autopsy. Among the remaining 14 cases, 12 patients underwent tissue sampling to confirm the initial diagnosis, including partial resection (two cases), open biopsy (eight cases), and computed tomography-guided biopsy (two cases); two patients (cases 13 and 14) harbored highly suggestive NMCs based on the MRI findings.

Recently, the association between aggressive fibromatosis and NMCs has been reported [1,9,13]. Hebert-Blouin MN et al. [9] reported that four patients had developed aggressive fibromatosis (three cases: sciatic nerve; one case: brachial plexus). The cause of aggressive fibromatosis is poorly understood. It is thought that manipulation during surgery or biopsy may be related to the development of aggressive fibromatosis in these lesions [3,9]. Ten of twelve patients who underwent tissue sampling for confirmation had post-biopsy follow-up MRI imaging. Of these 10 cases, six patients subsequently developed aggressive fibromatosis at the sites of previous manipulation. The MRI findings of aggressive fibromatosis manifested as lobulated masses with low to intermediated T1 and T2 signal intensities and strong enhancement [1]. Among them, three patients (cases 4, 6, and 8) had undergone re-resection, computed tomography-guided biopsy was performed in case 7, observation was performed in case 11, and there were no reports of further evaluation in case 12.

NMCs are known as benign tumors, but limited information is available regarding their diagnosis and treatment. The natural history and evolution over time of NMCs are also not well known. Among the reported cases, there were only 10 cases in which follow-up observations were reported. The median duration of follow-up was 2 years (range: 2 months-–8 years). Case 14 did not undergo tissue sampling after initial putative diagnosis, however.

the disease was noted to be stable until the 2-year follow-up. The three cases of partial resection (case 2) and open biopsy (cases 5 and 10) did not develop aggressive fibrosis on follow-up. Our patient has been monitored for 17 years without development of aggressive fibrosis, despite the patient having undergone surgical manipulation. It was revealed that the NMC was definitely a benign tumor, which had a slow-growing natural history. We presume that the extent of tumor growth may be related to the growth of the patient. In other words, this is the only case of long-term follow-up of an NMC. NMCs tend to be intermingled or closely associated with the parent sciatic nerve, which limits surgery. Surgery is very likely to cause potentially serious neurologic deficits. Markel F. at al reported the patient who had palsy of median and ulnar nerves after operation for neuromuscular choristoma [2,3]. In addition, the manipulation may lead to the development of aggressive fibromatosis. Considering the above explanations including long-term natural history, the diagnosis of NMCs is thought to be possible prior to biopsy or resection based on the unique and characteristic MRI findings with consistent clinical findings. Therefore, a diagnosis based on MRI findings should dissuade the clinician from proceeding with tissue confirmation.

4. Conclusion

NMCs demonstrate typical and distinctive MRI findings, which can result in a definite diagnosis in the clinical setting. Recent reports have revealed that biopsy or surgery for NMCs confer the potential for aggressive fibrosis. Therefore, if an NMC is suspected based on MRI findings, biopsy should be avoided.

Table 1
Literature review of sciatic nerve neuromuscular choristoma

Case	Age/sex	Site	Symptoms	MRI	Intervention	Fibromatosis		Follow-up	Ref.
						Onset	Treatment		
1	68/F	Left	Cavovarus foot Atrophy Weakness	None	Diagnosis at autopsy	None	None	None	[11]
2	0/M	Not known	Palpable mass	None	Partial resection	None	None	Stable for 3.5 years	[12]
3	2/M	Right	Limp Atrophy	None	Partial resection	Not known	Not known	No further FU mentioned	[2]
4	4/F	Left	Cavovarus foot Spasticity	None	Open biopsy	2 years	Resection	No further FU mentioned	[6]
5	8/M	Right	Cavovarus foot Limp	Soft-tissue mass parallel to the sciatic nerve	Open biopsy	None	None	Stable for 18 months	[7]
6	18/M	Right	Pain Weakness	Fusiform enlargement of the right sciatic nerve	Open biopsy	8 years	Resection Radiotherapy	Recurrence 8 months after resection	[1,8]
7	42/M	Left	Pain	Fusiform enlargement of the left sciatic nerve	CT-guided biopsy	3 years	CT-guided biopsy & Observation	After 2 years, 2nd biopsy, increased size	[9]
8	5/F	Right	Weakness Cavovarus foot	None	Open biopsy	4 months	Chemotherapy for 18 months & resection	Recurrence 9 months after resection	[9]
9	14/M	Right	Weakness	Large fusiform enlargement of right sciatic nerve	Open biopsy	Not known	Not known	No further FU mentioned	[1]
10	11/M	Right	Pain Weakness Shorter right leg	Well-circumscribed, mildly heterogeneously enhanced mass in the right sciatic nerve	Open biopsy	None	None	Stable for 1.5 years	[3]
11	11/M	Right	Pain Weakness Limp Shorter right leg	Homogeneous, mild enhancement of the right sciatic nerve	CT-guided biopsy	6 months	Observation	After 18 months, post-biopsy, increased size	[1]
12	10/F	Right	Shorter right leg	Fusiform enlargement of the right sciatic nerve	Open biopsy	2 months	Not known	No further FU mentioned	[10]
13	45/F	Left	Pain Shorter left leg	Fusiform enlargement of left sciatic nerve	None	Not known	Not known	No further FU mentioned	[1]
14	3/M	Right	Weakness Atrophy	Fusiform enlargement of the right sciatic nerve	None	None	None	Stable for 2 years	[4]
15 (our case)	0/F	Left	Palpable mass at birth (2001) Weakness (2006)	Fusiform enlargement of the left sciatic nerve	Open biopsy	None	None	Stable for 17 years	

MRI, magnetic resonance imaging; FU, follow-up; CT, computed tomography.

Acknowledgments

None.

Conflicts of interest

The authors declare that they have no conflict of interest.

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