Foot Drop as an Unusual Presentation of Plexiform Neurofibroma: A Case Report of an Otherwise Healthy Female

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ABSTRACT

Objectives: To describe an unusual presentation of plexiform neurofibroma in an otherwise healthy female patient.

Study design: Case report.

Setting: Department of Rehabilitation Medicine, King Chulalongkorn Memorial Hospital, Bangkok.

Subjects: A 46-year-old otherwise healthy female presenting with right foot drop.

Methods: Not applicable.

Results: An electrodiagnostic study was performed by a physiatrist to determine the cause and evaluate the severity of the foot drop and to reveal any evidence of abnormalities extending to the sacral plexus. The evidence discovered lead to further investigation, including magnetic resonance imaging (MRI) of the plexus, which confirmed the diagnosis of plexiform neurofibroma. The patient had no skin lesions and no family history of neurofibroma. The tumor was removed and a biopsy confirmed an intraneural neurofibroma. A plastic ankle foot orthosis was prescribed to assist ankle dorsiflexion and improve walking. Six months after surgery, power of the involved muscles showed no significant improvement.

Conclusions: Foot drop in healthy individuals can result from plexiform neurofibroma even without a definitive diagnosis of neurofibromatosis type 1. Electrodiagnostic study should be the first step before undertaking further investigation, e.g., MRI. MRI is prescribed to confirm the diagnosis and location of the lesion.

Keywords: foot drop, plexopathy, plexiform neurofibroma, electrodiagnosis, rehabilitation

ASEAN J Rehabil Med. 2022; 32(1): 44-47.

Introduction

Foot drop is common presentation leading patients to visit a physiatrist for a definite diagnosis and proper appropriate management. Foot drop can result from central, peripheral, and other metabolic causes. Injury at any point of along the peripheral nerve, especially a peroneal nerve injury, is the most common cause of foot drop.[1]

Electrodiagnostic study commonly involves an investigation to locate the lesion and to evaluate the cause and severity of the foot drop. Neurofibromas are benign nerve tumors that contain multiple nerve fascicles.[2] They are commonly poorly circumscribed and locally invasive.[3] Plexiform neurofibroma is generally believed to present exclusively in patients diagnosed with neurofibromatosis type I and is a pathognomonic sign.[3] There are few case reports of neurofibroma involving the lower extremities.[2, 4-6] For that reason, we report this rare case of an otherwise healthy female presenting with foot drop which was later diagnosed as plexiform neurofibroma.

Case presentation

A 46-year-old female presented with right foot drop at the rehabilitation out-patient department. She recognized the foot drop in her right foot from difficulty climbing stairs and frequent falls for a year before this visit. She also had numbness at the anterolateral part of her right leg and the dorsum of her right foot. She denied any underlying disease or traumatic events. She had history of frequent alcohol drinking while sitting crossed-legged. Right foot drop with steppage gait, muscular atrophy at both anterior and posterior aspects of the right lower leg as well as the posterior thigh were observed. There were no skin lesions or brown freckling spots (cafe-au-lait macules), brown dome shape lesions (neurofibromas) or brown pigmentation in the eye (Lisch nodules). The pin-prick sensation was diminished at the anterolateral aspect of the right leg and dorsum of the right foot. Muscle power of the upper extremities was normal but some muscles of the lower extremities were severely weak or paralyzed as shown in Table 1. Tinel's sign was positive at the right fibular head area with a tingling sensation down to the right foot. Deep tendon reflexes (DTRs) were 1+ for all extremities. Electrodiagnostic study was performed by a physiatrist.

An electrodiagnostic study, including a nerve conduction study and needle EMG, were performed and the findings are shown in Tables 2, 3 and 4. The diagnosis was incomplete right L5-S1 sacral plexopathy with signs of chronic degeneration at the L4-5 level and active degeneration at the S1 level. Further investigation of cause of the sacral plexopathy was suggested.

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Received: 19th August 2021 Revised: 22th September 2021 Accepted: 28th September 2021

Table 1. Muscle power of the lower extremities according to the Medical Research Council (MRC) grading

Muscles	Hip flexors	Hip extensors	Knee flexors	Knee extensors	Hip abductors	Ankle dorsiflexors	Ankle plantar flexors	Tibialis posterior	Peroneus longus	Extensor hallucis longus
Right	5	3	5	5	5	1	5	5	1	0
Left	5	5	5	5	5	5	5	5	5	5

Table 2. Findings and interpretation of sensory nerve conduction study

Nerve - site of stimulation	Onset latency (ms)	Peak amplitude (microV)	Peak-Peak amplitude (microV)	Duration (ms)	Distance (cm)	Velocity (m/s)
Sural nerve - lateral malleolus						
Left	2.4	18.4	15.7	2.03	10	40.9
Right	2.6	9.7	6.6	1.93	10	38.4
Superficial peroneal						
Left	2.7	6	5.1	2.45	12	44.3
Right						
Saphenous - Ankle						
Left	2.8	6.1	1.5	1.56	12	43.5
Right	2.7	5.1	0.59	1.30	11	41.4

Interpretation of sensory nerve conduction study	
Right sural nerve	Low SNAP amplitude, normal latency and velocity
Right superficial peroneal nerve	No response
Right saphenous nerve	Normal latency, amplitude and velocity
Left sural nerve	Normal latency, amplitude and velocity
Left superficial peroneal nerve	Normal latency, amplitude and velocity
Left saphenous nerve	Normal latency, amplitude and velocity

SNAP, sensory nerve action potential; ms, millisecond, microV, microvolt; cm, centimeter; m/s, meter/second

Table 3. Findings and interpretation of sensory nerve conduction study

Nerve - site	Latency (ms)	Amplitude (mV)	Distance (cm)	Velocity (m/s)
Left Common peroneal nerve - EDB	5	6.2		
	12.66	5.4	35	45.7
Right Common peroneal never - EDB	7.55	0.1		
	19.74	0.1	30	24.6
Left Tibial nerve - AH	3.54	21.1		
	11.77	15.1	38	46.2
Right Tibial nerve - AH	3.96	19.4		
	13.07	15.4	36.5	40

Interpretation of motor nerve conduction study	
Right common peroneal nerve	Low CMAP amplitude, prolonged latency and slow velocity
Right tibial nerve	Normal latency, amplitude and velocity
Left common peroneal nerve	Normal latency, amplitude and velocity
Left tibial nerve	Normal latency, amplitude and velocity

CMAP, compound muscle action potential; ms, millisecond; mV, millivolt; cm, centimeter; m/s, meter/second; EDB, extensor digitorum brevis muscle; AH, abductor hallucis muscle

When the electrodiagnostic findings suggested plexopathy, the physiatrist requested a magnetic resonance imaging (MRI) of the sacral plexus to identify and locate any lesions of the lumbar and sacral plexus. The MRI showed a 1.1x3.4x4.5 cm ill-defined elongated shape heterogenous enhancing lesion located anteriorly to the right sacral alar and extending into the right sciatic foramen along the right L5 nerve root, probably a plexiform neurofibroma (Figure 1). In addition, it revealed lumbar spondylosis with degenerative discs and central disc herniations causing stretching of the bilateral L5 traversing nerve roots (predominantly on the left) and an extraforaminal left L5 exiting nerve root. At this stage, a neurosurgeon and a neurologist were consulted to do a workup on neurofibromatosis and treatment of plexiform neurofibromas.

Table 4. Needle EMG summary table

Muscles of the right lower extremity	Insertional activity	Fib	PSW	Amp	Dur	PPP	Recruitment pattern
Rectus femoris	Normal	None	None	Normal	Large	Normal	Normal
Tibialis anterior	Increased	2+	2+	Normal	Normal	Normal	Discrete
Gastrocnemius (medial head)	Normal	None	None	Normal	Normal	Normal	Normal
Peroneus longus	Increased	2+	2+	Normal	Normal	Normal	Discrete
Tibialis posterior	Normal	None	None	Normal	Normal	Normal	Reduced
Biceps femoris (short head)	Normal	None	None	Normal	Normal	Normal	Reduced
Gluteus maximus	Increased	2+	2+	Normal	Normal	2+	Reduced
Lumbar paraspinal muscles (L5-S1)	Increased	None	None	Normal	Normal	Normal	N/A*

Abbreviations: EMG, electromyography; MUAP, motor unit action potential; Fib, fibrillation potential, PSW, positive sharp wave; Amp, amplitude; Dur, Duration; PPP, polyphasic potential; N/A, not applicable

* Recruitment pattern was not evaluated at paraspinal muscles

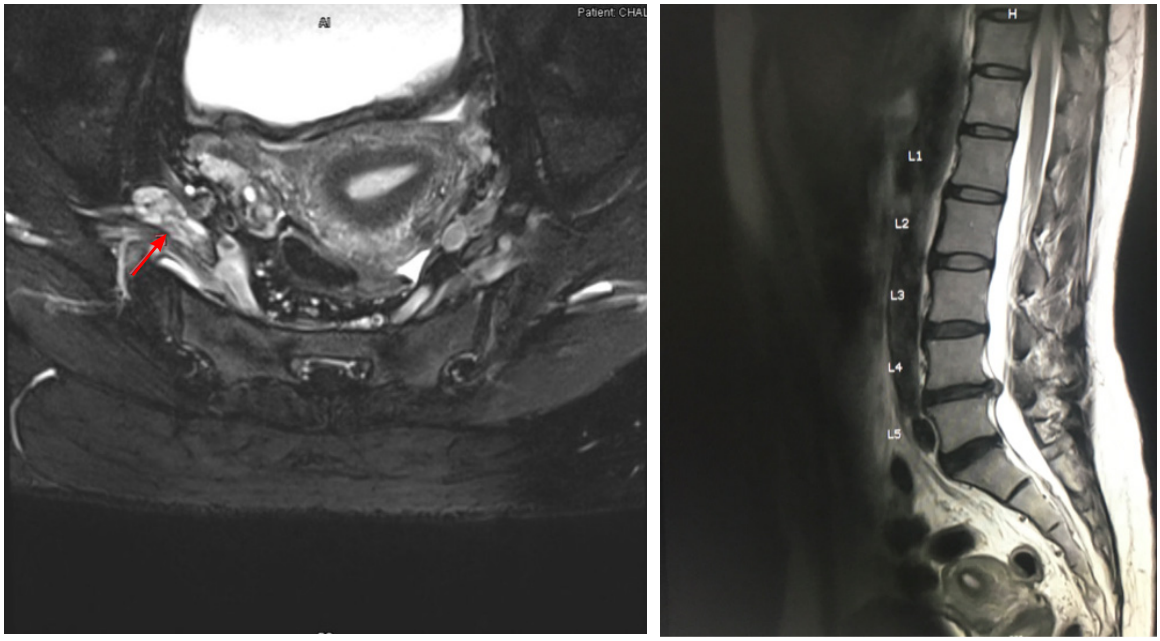

Figure 1. Magnetic resonance imagining (MRI) of the sacral plexus. The red arrow points to a mass located anteriorly to the right sacral alar

While waiting for surgery, the physiatrist prescribed an outpatient rehabilitation program which consisted of range of motion and strengthening exercises and electrical stimulation (ES). In addition, the patient was instructed to apply home-use ES at the weak/paralyzed muscles once or twice a day. A plastic ankle foot orthosis was also prescribed so the patient could ambulate comfortably.

The surgery, a transabdominal retroperitoneal approach for tumor removal, was done by a neurosurgeon. Pathology examination of the mass confirmed the diagnosis of intraneural neurofibroma. There were no changes in muscle power after the surgery.

A follow-up MRI at 6 months after surgery revealed no enhanced neurofibroma of the right L5 or other lumbar nerve roots, indicating probable total removal of the tumor; however, There was no neurological improvement after surgery and the patient continued to complained of fatigue after walking long distances.

Discussion

This is a rare case of an otherwise healthy woman presenting with foot drop due to plexiform neurofibroma without a diagnosis of neurofibromatosis type I. Neurofibromatosis type I (NF type I) was first described by Frederich von Recklinghausen in 1882.[7] NF type I is a relatively common neurocutaneous disease with an autosomal-dominant inheritance pattern.[4] About 50% of individuals with NF type I have no family history of the disease is, indicating the disease is due to de novo mutations in the 17.q11.2 chromosome. Diagnostic criteria of neurofibromatosis were established by the National Institutes of Health (NIH) Consensus Development Conference on Neurofibromatosis in 1998 and are shown in Table 5. In this case, the patient met only one criterion, a single plexiform neurofibroma, which was not compatible with the NIH criteria for a definitive diagnosis of neurofibromatosis type 1. Plexiform neurofibroma is one of the non-malignant features that can also be found in patients with NF type I.[4,7,8]

Table 5. The National Institutes of Health (NIH) criteria for neurofibromatosis type 1 (From the NIH consensus development conference 1998)[7]

Clinical diagnosis based on presence of two of the following:
1. Six or more café-au-lait macules over 5 mm in diameter in prepubertal individuals and over 15 mm in greatest diameter in post pubertal individuals.
2. Two or more neurofibromas of any type or one plexiform neurofibroma.
3. Freckling in the axillary or inguinal regions.
4. Two or more Lisch nodules (iris hamartomas).
5. Optic glioma.
6. A distinctive osseous lesion such as sphenoid dysplasia or thinning of the long bone cortex, with or without pseudarthrosis.
7. First-degree relative (parent, sibling, or offspring) with NF-1 by the above criteria

Neurofibromas may occur anywhere along a nerve from the dorsal root ganglion to the terminal nerve branches.[2] Plexiform neurofibroma without neurofibromatosis type I is uncommon and rarely presents in combination with foot drop.[4] There are few case reports published about neurofibroma involving the lower extremity. In 1994, Nagel et al,[5] reported on an athletic patient who presented with a non-traumatic peroneal neuropathy which failed to resolve after a period of rest. In that case, finally imaging showed a multilobulated mass along the course of the common peroneal nerve consistent with a plexiform neurofibroma. Another report described the case of a patient who suffered from chronic anterior leg pain that failed to respond to medical treatment, including multiple injections.[6] In that case, electromyography identified abnormal findings in the accessory peroneal nerve branch in the discomfort area. Finally, MRI results showed a hyperintensity signal and perifascial hyperintensity superficial to the anterior musculature with a nodular-like signal measuring about 6 mm.

A review of the literature found that the presenting symptoms usually include pain, soreness or a tingling sensation at the tumor area as well as weakness in the legs and difficulty walking or running. Chang et al,[2] reported a case of neurofibromatosis type 1 with a painful palpable mass, about 10 cm in diameter, in the right leg for 1 year. MRI revealed low-to-intermediate signal intensity between the anterior tibia, peroneus longus, and brevis muscles on T1-weighted images (T1WI), and high signal intensity on T2-weighted images (T2WI). The T2WI images were characterized by a low-density center and a hyperintense rim. After a tumor removal operation, the patient's soreness, tingling sensation, and tolerance of long periods of standing improved dramatically.

Treatment of plexiform neurofibromas is usually conservative owing to the high rate of recurrence and significant morbidity associated with surgical resection requiring sacrifice of the parent nerve.[3,4,7,9] In the present case, the patient was operated on and the tumor seems to have been totally excised without further weakness but with no recovery of strength. This might be due to the patient having been seen late, one year after the onset and when, prior to the surgery, the involved muscles had become completely or nearly completely paralyzed. At 6 months after surgery, power of the involved muscles still had not changed. ES seemed not to provide benefit in this case due to the severe muscular atrophy, but it did irritate the patient's skin. Plastic AFO, range of motion and strengthening exercises of the muscles around the hip and ankle were beneficial. The patient could walk safely and comfortably with a plastic AFO in her daily activities.

Conclusions

This study presents a rare case of a woman presenting with foot drop without any underlying disease. A physiatrist provided comprehensive care, including complete history taking, physical examination, an electrodiagnostic study and proper rehabilitation treatment. After the electrodiagnostic study, further imaging was indicated which revealed the true cause of foot drop.

References

1. Carolus AE, Becker M, Cuny J, Smektala R, Schmieder K, Brenke C. The interdisciplinary management of foot drop. Dtsch Arztebl Int. 2019;116:347-54.
2. Chang LR, Shieh SJ. Neurofibroma derived from the deep peroneal nerve: a case report. Kaohsiung J Med Sci. 2006;22:290-6.
3. Lin V, Daniel S, Forte V. Is a plexiform neurofibroma pathognomonic of neurofibromatosis type I? Laryngoscope. 2004;114:1410-4.
4. Atkins NK, Stensby JD, Gaballah AH. Lumbosacral plexiform neurofibroma: a rare case in an adult without neurofibromatosis type I. Skeletal Radiol. 2020;49:321-30.
5. Nagel A, Greenebaum E, Singson RD, Rosenwasser MP, McCann PD. Foot drop in a long-distance runner. An unusual presentation of neurofibromatosis. Orthop Rev. 1994;23:526-30.
6. Carpenter B. Neurofibroma of the anterior aeg: a case report. FAOJ. 2012;5. doi: 10.3827/faoj.2012.0506.0002
7. Hirbe AC, Gutmann DH. Neurofibromatosis type 1: a multidisciplinary approach to care. Lancet Neurol. 2014;13:834-43.
8. Tadini G, Milani D, Menni F, Pezzani L, Sabatini C, Esposito S. Is it time to change the neurofibromatosis 1 diagnostic criteria? Eur J Intern Med. 2014;25:506-10.
9. Ferner RE. Neurofibromatosis 1 and neurofibromatosis 2: a twenty first century perspective. Lancet Neurol. 2007;6:340-51.