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Atypical presentation of syringomyelia mimicking as motor neuron disease: A case report

Zenyros Faith A Sabellano*; Laurence Kristoffer J Batino; Jose C Navarro

Jose R Reyes Memorial Medical Center, Department of Neurology, Manila, Philippines.

***Corresponding Author: Zenyros FA Sabellano**
Jose R Reyes Memorial Medical Center, Department of Neurology, Manila, Philippines.
Email: zfsabellano@gmail.com

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Abstract

This is a case involving a 27-year-old Filipino female who presented with a chief complaint of progressive weakness in all extremities. The initial assessment of the patient's clinical history and the neurologic examination revealed the presence of a motor neuron disease. This conclusion was drawn based on the observation of both upper and lower motor neuron signs. Furthermore, the findings from the Electromyography (EMG) Nerve Conduction Velocity (NCV) findings suggest active and chronic generalized disorder of motor neurons. However, craniospinal MRI revealed the presence of a long segment C1-T12 syringomyelia, accompanied by an extradural, extramedullary cystic mass at the T12 to L1 level. This paper is the first one to report an atypical presentation of syringomyelia as a motor neuron disease secondary to a non-chiari malformation.

Keywords: Atypical syringomyelia; Motor neuron disease.

Introduction

A 27-year-old Filipino female presented to the outpatient department with complaints of weakness of all extremities that began in July 2016. Initially, she experienced proximal weakness in her right upper extremity, with difficulty raising her arm above shoulder level. There were no associated symptoms or neurologic deficit. Over time, the weakness progressed distally resulting in poor grip that interfered with activities such as writing and using utensils. Eventually, she started dropping objects from her right hand. There were no other accompanying symptoms such as numbness, paresthesia, and weakness on the left extremities and right lower extremity. Consequently, she decided to seek consultation at a nearby hospital, where a plain cranial CT scan showed no notable findings.

In 2021, she began experiencing left upper extremity weakness characterized as heaviness and poor grip, now affecting both upper extremities. Hence, an inability to use her hands for activities of daily living, such as feeding, bathing, and changing clothes. She occasionally noticed twitching of muscles in her right arm, which occurred at no particular time of day or during any specific activity. In the second quarter of 2022, she noted weakness in her right lower extremity, making it difficult for her to put on slippers. There was no numbness, paresthesia, or back pain reported. She also observed spasticity in both upper extremities, contractures in both hands, and muscle atrophy. As a result, she required assistance with all activities of daily living. In the second week of January 2023, there was further progression of weakness, affecting the distal part of her left lower extremity. However, she was still able to walk without assistance. There were no other accompanying symptoms such as numbness, bowel and bladder problems, nape pain, back pain, or tingling sensation.

The patient's past medical history revealed no history of trauma, previous infections, or hospital admissions. She was born to a G5P5 mother without any birth complications. There is a family history of hypertension but no history of progressive motor weakness, pulmonary diseases, thyroid diseases, or malignancy. The patient does not smoke, drink alcoholic beverages, or use illicit drugs. She denies exposure to fertilizers, toxins, and heavy metals. There has been no recent local or in-

ternational travel. In terms of her sexual history, she had her menarche at the age of 14 years and has regular menstruation lasting 5-7 days, requiring 2-3 pads per day, with no reported dysmenorrhea or discomfort. She is nulligravid and denies a history of coitarche.

During the general physical examination, muscle atrophy and contractures were observed in both upper extremities, particularly in both hands. Palpation of her back revealed no tenderness or palpable masses. On neurological examination, the patient did not present with any cranial nerve deficits but exhibited predominant weakness in both upper extremities and the proximal part of the right lower extremity (Table 1). There was no spinal level or numbness reported in all extremities, including her upper back, upon light touch and pin prick tests. Proprioception, temperature sensation, and vibration sensation were intact in all extremities. The patient also displayed fasciculations in her right upper extremity, spasticity in all extremities, increased muscle tone, and bilateral ankle clonus. Superficial reflexes were absent and there were no meningeal signs. Presence of bilateral Babinski sign and Hoffman sign on the right were elicited. Cerebellar signs in the upper extremities could not be assessed due to contractures; however, the patient was able to perform bilateral heel-to-shin testing. During gait and stance examination, the patient exhibited a narrow base with knees and thighs touching midline, with a spastic gait.

Table 1: Motor examination on all extremities.

Upper extremities		Right	Left	Lower extremities		Right	Left
Shoulder	Abduction	2/5	2/5	Hips	Abduction	5/5	5/5
	Adduction	2/5	1/5		Adduction	5/5	5/5
	Flexion	2/5	2/5		Flexion	5/5	5/5
	Extension	0/5	0/5		Extension	5/5	5/5
Elbow	Flexion	3/5	3/5	Knee	Flexion	4/5	5/5
	Extension	2/5	4/5		Extension	4/5	5/5
Wrist	Flexion	3/5	3/5	Ankle	Dorsiflexion	5/5	5/5
	Extension	4/5	3/5		Plantarflexion	5/5	5/5
Finger	Abduction	1/5	1/5		Inversion	5/5	5/5
	Adduction	1/5	2/5		Eversion	5/5	5/5
	Flexion	3/5	3/5				
	Extension	0/5	0/5				

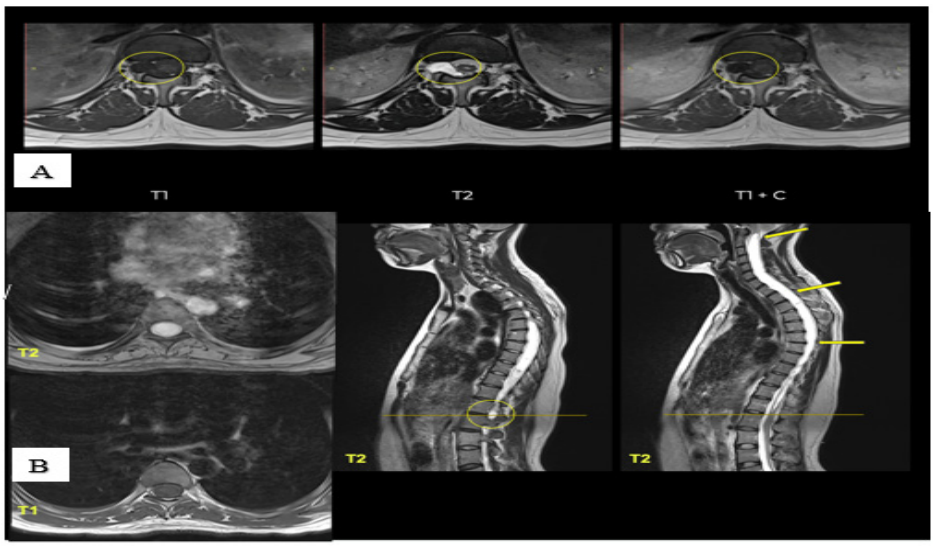

Figure 1A: Axial view shows an extramedullary extradural T1 hypointense and T2 hyperintense focus at the right side of the vertebral foramen extending into the right neural foramina, measuring 1.4 x 1.7 x 3.2 cm. It exhibits no enhancement on post-contrast studies.

Figure 1B: Shows the more proximal central canal of the spinal cord is dilated up to the level of the foramen magnum. The dilated spinal canal appears as a centrally located intramedullary elongated/tubular structure of high T2W and low T1W signal intensity. There is expansion of the involved spinal cord-widest at C7 level measuring 1.7 cm in its widest cross sectional diameter and 2.2 cm in its widest transverse diameter. There is no evidence of contrast enhancing intraspinal mass or abnormal enhancement.

Table 2: Summary of EMG NCV findings.

				EMG findings	Summary		
Muscle	Side	Insertion	FIBS	Sharp Wave	FASC	Duration	Recruitment
1st Dorsal Inter.	L	Normal	None	None	None	Normal	Decreased
Biceps Brachii	L	Normal	None	None	None	Normal	Decreased
Gastroc.Medial H	L	Normal	None	None	None	Normal	Decreased
Tibialis Anterior	L	Normal	None	None	None	Normal	Decreased
1st Dorsal Inter.	R	Normal	None	None	None	Normal	Decreased
Biceps Brachii	R	Normal	+1	+1	+1	Normal	Decreased
Gastroc.Medial H	R	Normal	None	None	None	Normal	Decreased
Tibialis Anterior	R	Normal	None	None	None	Normal	Decreased
Geniohyoid	N	Normal	+1	+1	+1	Normal	Not activated
Paraspinal, Cervical	N	Normal	None	None	None	Normal	Not activated

Diagnostics were the following: Electromyography (EMG) and Nerve Conduction Velocity (NCV) showed evidence of active and chronic generalized disorder of motor neurons (refer to Table 2). Craniospinal MRI with contrast was done which only revealed mildly dilated ventricular system with no adjacent periventricular CSF seepage indicative of communicating hydrocephalus. This may be possibly related to the central canal dilatation seen on the cervical cord. There was no evidence of chiari malformation. Contrast enhanced whole spine MRI showed an extradural, extramedullary cystic lesion at the right side of the vertebral foramina that is closely associated with the right T12-L1 exiting nerve roots causing mass effect with considerations of arachnoid or perineural cyst, spinal canal stenosis and subsequent C1-T12 long segment syrinx (See Figure 1).

Discussion

Syringomyelia is a neurological disorder in which a fluid-filled cyst (syrinx) forms within the spinal cord. The Cerebrospinal Fluid (CSF), which surrounds and protects the spinal cord and the brain- builds up within the tissue of the spinal cord, expanding the central canal, hence also termed as central cord syndrome. Patients typically present with a combination of lower motor neuron signs at the segmental level, a dissociation sensory loss in a cape-like pattern in the arms and upper trunk distribution [1]. There are many several causes of syringomyelia and it can therefore be of two major forms [2]: (A) congenital syringomyelia secondary to chiari malformation, which is the most common among all cases; and (B) acquired syringomyelia which is secondary to spinal cord injury, meningitis, arachnoiditis, and spinal cord tumor.

Bogdanov [3] et al. conducted a study with the aim of investigating whether syringomyelia associated with chiari malformation and syringomyelia unrelated to chiari malformation presented distinct symptoms. The results from their series demonstrated that the occurrence of symptoms was similar in both groups. The most commonly reported symptom was segmental sensory loss, which was observed in 93% of cases, followed by the presence of pyramidal signs (82%) and muscle atrophy (60%) [3].

The diagnosis of syringomyelia should always be based on clinical suspicion. Despite the typical clinical neurologic diagnosis of syringomyelia as a central cord syndrome, atypical presentations have been reported [1]. The reported atypical presentations were themed into seven types, total of 23 case reports: (A) symptoms of peripheral neuropathy, (B) limb dystrophy, (C) limb hypertrophy, (D) movement disorder, (E) brainstem syndrome, (F) neuroarthropathy, (G) non specific clinical presentation. There is one reported case of an amyotropic lateral sclerosis mimic due to chiari malformation and syringomyelia. This paper is the first one to report an atypical presentation of syringomyelia as a motor neuron disease secondary to a non-chiari malformation [4-6].

Syringomyelia associated with a spinal arachnoid cyst was also studied as a cause of syringomyelia, however is very unusual. The annual incidence of syringomyelia in patients with an arachnoid cyst is known to be rare, with only eight reports known [4]. The origin of the cysts is caused by the dilatation of the septum posticum, a thin membranous arachnoid partition that longitudinally divides the posterior subarachnoid space in the midline explaining the posterior location of the cysts. The presence of an arachnoid cysts can cause numbness (42%), motor disturbances (40%), and pain (23%). Our patient did not manifest any numbness and pain, however initially presented with right upper extremity weakness which progressed to affect the other extremities.

As syringomyelia is caused by the obstruction of cerebrospinal fluid (CSF) flow through the spinal subarachnoid space, the obstruction can be classified as follows [5]: (A) Complete obstruction wherein CSF pressure pulse wave reaches its blocking point, and is transmitted to the spinal cord. (B) Partial obstruction which allows some flow of the CSF, which can be explained by two principles. The first is an increase in CSF flow speed due to narrowness of the channel, resulting in decreased pressure in that region, known as Bernoulli's principle. The second phenomenon is the Venturi effect, wherein a fluid flowing at a high speed creates a suction effect. Both principles result in the dilatation of the parenchyma below the obstruction. Once the parenchyma is dilated, the extracellular space increases, allowing CSF accumulation and progressive increase in the cavity size-leading to the formation of syrinx. Considering the craniospinal MRI of the patient, a partial obstruction can be deduced due to the presence of communicating hydrocephalus.

The patient underwent symptomatic surgery through suboccipital craniectomy with C4 laminectomy and syringo-subarachnoid shunt insertion at C4, which focuses on relieving the symptoms. Following the operation, the patient demonstrated postoperative improvement in motor weakness. Post operative findings showed syrinx with clear CSF. Few days after the surgery, she had the ability to lift both upper extremities below shoulder level, and there were no motor deficits observed in the lower extremities. Additionally, she regained hand functionality, allowing her to grip objects and use her cellular phone.

Conclusion

This is the first reported case of syringomyelia, resembling a motor neuron disease, characterized by the presence of both upper and lower motor neuron signs during neurological examination, as confirmed by EMG NCV. Importantly, the patient's medical history and clinical neurologic assessment did not indicate a central cord syndrome, which is the typical presentation for syringomyelia. The craniospinal MRI scan with contrast then revealed the co-existence of an arachnoid cyst at the right vertebral foramen level. The cyst could potentially be responsible for the development of a substantial syrinx. The patient did not, as well, presented with pain or numbness which can support the diagnosis of an arachnoid cyst.

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