Rare presentation of lower limb weakness, a case report

Hamzeh Yousef khmour
Salma el Nour
Mansour Elmahdi

Specialist Family Medicine, Qatar

Correspondence:
Dr. Hamzeh Yousef khmour, LPI
Specialist Family medicine,
Qatar
Tel: +974 33304316
Email: hkhmour@phcc.gov.qa, hkhmour@gmail.com

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Abstract

SUMMARY
This is a 53 year old male, who was previously healthy, and who presented to a primary care physician, complaining of from chronic back pain. He was seen multiple times with the same problem, which was treated as Myalgia with pain killers only. After a proper approach and investigation he was found to have ALS Amyotrophic lateral sclerosis.

BACKGROUND:
Lower limb weakness is a common problem in the primary care setting, with a multiple differential diagnosis. It is defined as loss of muscle strength[1]. The physician should firstly isolate and be sure that the complaint of patient is weakness of the muscle , mainly by investigating other causes that may mimic and overlap with the presentation; the famous and most common cause being fatigue[2]. History and physical examination in addition to investigation will assure that the physician will reach the proper diagnosis and management plan but before that knowing the mechanisms of weakness is the corner stone in approach[3,5], which includes[3,4]:

Upper motor neurons, Lower motor neurons, Neuromuscular junction, Muscle.

In the primary care setting and because of patient flow, usually common is common, which is exactly what happened to the patient, as he visited the center multiple time because of weakness and mild pain which did not attract the attention of the treating physicians to more serious and complicated disease[6,7].

Amyotrophic lateral sclerosis (ALS), commonly called Lou Gehrig’s disease, is a progressive neurodegenerative disease affecting both upper and lower motor neurons. ALS is a condition characterized by weakness, muscle wasting, fasciculations and increased reflexes[1,2]. The annual incidence rate is one to three cases per 100,000. The disease is mostly diagnosed in middle age and affects more men than women [5].

Over a period of months or years, patients with ALS develop severe, progressive muscular weakness and other symptoms caused by loss of function in both upper and lower motor neurons.

Mainly it presents with 2 major categories of symptoms: Upper motor neuron, Lower motor neuron [2].

In the patient’s case it was mainly lower motor neuron symptoms without any symptoms of upper motor which makes the presentation unique and confusing with other differential diagnoses.

ALS has a bad prognosis and limited options of treatment. Respiratory failure limits survival to 2-5 years after disease onset. RILUZOLE is the only drug that can affect prognosis [1,7,8,9].

Being rare, difficult to treat and with a bad prognosis, makes us want to share this case with our colleagues as the learning opportunity in this case makes it a valuable one for a case report.

Key words: lower limb weakness, case report
Case Presentation

A 53 year old male working as a driver, was known to have recurrent chronic back pain for one year, that became progressive in the last 3 months. The pain was associated with weakness of both lower limbs and inability to walk without assistance.

He visited the health center multiple times to get a diclofenac injection which he believed helped in relieving his pain and improving the limb weakness. His condition became worse and not improving on the painkiller. He could not walk without assistance, depending on a cane for walking for a period. Weakness progressed rapidly until he became wheelchair bound. No other neurological or sensory symptoms were associated with lower limb weakness apart from the inability to hold urine. There was no fever, no weight loss, no history of trauma. Other System review was unremarkable.

His Past medical history: Patient report using pregabalin for neuropathic pain back in his country, Family history is insignificant, no similar condition in the family, no history of neurological disease, worked as a driver since 10 years in Doha, and he is a smoker of 25 packs per year, not alcoholic (last drink according to him was many years ago) and on regular diet not vegan or vegetarian

He has a major concern regarding his pain as it is preventing him from driving in the past 3 months.

Examination at presentation to health center:
Patient presented in wheelchair, he was not in pain, vitally stable and looked well, his chest was clear, with normal s1 s2 sounds.

We focused on neurological and lower/upper limb examination at that time, cranial nerve examination was unremarkable. When we examined the gait it showed that he has a picture of left foot drop. The Upper limbs showed Weakness on Right distal muscles, No proximal muscles weakness. And in Lower limbs Motor Tone was Decreased, Power 3/5 throughout. Proximal muscles, 1/5 on dorsiflexion and plantar flexion. Sensory: intact to light touch and pain throughout in all limbs. Reflexes examination showed Normal Knee reflexes, Abnormal Ankle reflexes, Plantar reflex was Downward on right side and Equivocal on left side, with positive Romberg sign. And on back examination there was No tender point, negative Straight leg raise test, Intact perineum sensation.

Investigations (If relevant)
Investigation at Primary care:
-XR Lumbar spine
Plain X-Ray Of The Lumbo-sacral Spine Revealed: -(report of x ray)

“Straightened lumbar lordosis…. spasm. Minimal Spondylo-degenerative changes of lumbar vertebrae showing anterior osteophytes at their opposing vertebral end plates. Minimal narrowing of L5/S1 disc space….. ?degenerative, ?variant. Intact both sacro-iliac joints.”

Investigations at secondary Hospital:
Patient was referred to ED for further evaluation and management.
MRI was done:
-MRI Spine thoracic lumbar with contrast (Verified)

“CLINICAL INDICATION: 53-year-old male patient. Examination was initially requested by neurosurgery with a clinical history of bilateral lower limb weakness and L1 Para spinal tenderness.

COMPARISON: No previous imaging available for comparison.

FINDINGS:
On screening imaging there is satisfactory alignment of the cervical spine. The vertebral body heights are well-maintained. There is degenerative disc disease at the level of C5-C6 with mild posterior osteophyte formation and mild disc bulge. There is also some degenerative disease at the level of C6-C7 with posterior disc bulge which is compressing the thecal sac.

No spinal cord lesions identified.

There is satisfactory alignment of the thoracic spine. The vertebral body heights are well contained. No fracture identified. There is small posterior disc bulge at the level of T4-T5.

There is satisfactory alignment of the lumbar spine. The vertebral body heights are well maintained. No fracture identified.

L4-L5: Mild diffuse disc bulge which is indenting the thecal sac and is abutting the traversing left L5 nerve root.

There is also mild-to-moderate bilateral foraminal narrowing. There is also bilateral facet joint effusion at this level.

L5-S1: There is diffuse disc herniation with a small central protrusion component which is abutting the right traversing S1 nerve root and abutting the left traversing S1 nerve root. There is also mild right foraminal narrowing and moderate left foraminal narrowing with abutment of the exiting left L5 nerve root.

IMPRESSION:
Multilevel degenerative disease as described above which is more significant at the level of L5-S1." Blood test in ER:
Myoglobin: 86 , CRP:6.4 , Urine analysis: -ve -Vasculitis and Gammopathy Screening:
ANA, CTD, ANCA, C3 , IgE , IgA, IgM ,IgG All are -ve -EMG and NCS (13/06/2021)

“Electrophysiologic evidence of active and generalized disorder of motor neurons and/or their axons consistent with amyotrophic lateral sclerosis.”
Differential Diagnosis: Sorted according to the presentation of case before the definitive diagnosis test

- Stroke
- Nerve Compression or Brain injury: neoplasm
- Guillain Barre syndrome
- Multiple Sclerosis
- Myopathy
- B12 deficiency.
- Alcoholic neuropathy.
- Infectious: HIV, Syphilis, Polio
- Spinal cord Injury
- Degenerative disc disease
- Cervical spondylosis
- Amyotrophic lateral sclerosis

Treatment and Outcome

The patient's only treatment was to solve the urine incontinence issue, started on:

- vitamin A/vitamin E
- Tolterodine: 4 mg, Oral, Daily

Started on physical therapy

Patient was diagnosed with ALS, followed and treated by Neurological department in HMC, started on above mentioned treatment and physiotherapy

Stable with regular follow up with Neurology and Respiratory

Further detail of treatment is not clear on the system and RILUZOLE treatment is not started yet.

Discussion

Lower limb weakness is not an uncommon presentation in primary care. A proper clinical systematic approach is essential to differentiate possible causes of lower limb weakness, as there is a wide possible differential diagnosis. Most important to exclude red flags for serious conditions if detected by primary care provider the patient should be immediately referred to emergency for further investigation and management.

Although ALS is a rare disease, and not presented frequently in primary care, it is a serious condition with a bad prognosis so early detection is essential.

When we approach the case at the beginning ALS was not on the top of our differential diagnosis; on the other hand, we were able to suspect a possibility of a serious underlying condition, even the patient himself was surprised and questioning why we are taking a full history and examining him.

We are not suggesting that every lower limb weakness is ALS or a similar condition. We recommend that primary care providers always keep in mind the following questions: what the lower limb weakness is, how to approach a patient with lower limb weakness and, what are the possible differential diagnoses.

Unfortunately, our patient was seen by more than one doctor in the health center and was misdiagnosed as a case of fatigue, or lower back pain as mentioned in his documents. Not only the risk of wrongly prescribed unnecessary treatments but also the risk of progression of the condition could be avoided in this case.

References

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