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Case Report

Friedreich's Ataxia Misdiagnosed as Lumbar Disc Prolapse: Case Report

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ABSTRACT

53-y-old veteran man without previous past medical history, suffered from severe low back pain, misdiagnosed as lumbar disc prolapse.

Keywords: Friedreich's ataxia; Back pain

INTRODUCTION

Friedreich's ataxia is the most common autosomal recessive spinocerebellar ataxia with variable pattern of presentations.

CASE DESCRIPTION

53-y-old veteran man with no past medical history presented with low back pain x 3y radiating to both LE occasionally with numbness of both feet and frequent falls and knee buckles. Was diagnosed as lumbar disc prolapse after positive MRI L5-S1 disc bulge, treated with physical therapy x 1 y and analgesics without improvement. Pain is 8-9/10, sharp exaggerated by prolonged standing and walking. Clinically: Lumbar lordosis, marked paravertebral muscle spasm, ROM: intact flexion, limited extension and side bending. FABERE: bilaterally positive. Neurologically: Alert, oriented x 3, normal speech and articulation, no nystagmus. Glove and stocking hypoesthesia. DTJ patellar G2, ankle G0 bilateral, UE jerks G2 bilaterally. MMT full UE and LE muscles except for R ankle dorsiflexor G4/5. Finger nose test and finger- finger test impaired. Heel-knee-shin test impaired. Romberg's sign positive and wide-based ataxic gait with partial R foot drop.

NCS and EMG UE and LE revealed axonal-demyelinating sensory polyneuropathy affecting all extremities.

DISCUSSION

This case represents Friedreich's ataxia misdiagnosed as lumbar disc prolapse based on MRI findings and chronic LBP, but the presence of sensory ataxia resulting in frequent falls and lack of balance were not detected and truncal ataxia with marked paravertebral muscle spasm could explain chronic back pain. Although MRI was positive for L5-S1 disc prolapse but this could not be correlated with clinical picture. NCS has proven the presence of sensory polyneuropathy which is a common feature of Fredrich's ataxia.

Revising literature regarding similar presentation of Friedreich's ataxia, we did not find similar presentation in term of the age, as the onset of Fredrich's ataxia is usually in younger age group than this current patient. This patient in particular was a very muscular healthy veteran. In addition to late onset presentation, the presenting feature which is low back pain was not listed in the literature among the common presenting feature of that disease, but one of the listed features is scoliosis which was not present in this particular patient.

Parkinson et al. reported the usual presentations of Friedreich's ataxia as; gait and limb ataxia, poor balance and coordination, leg weakness, sensory loss, areflexia, impaired walking, dysarthria, dysphagia, eye movement abnormalities, scoliosis, cardiomyopathy [1].

Regarding this particular patient, he had balance and gait disorder but was not the presenting feature. This may be compensated by his strong body musculature.

Reetz et al. reported that the most frequent presentation of Friedreich's ataxia was abnormal eye movement (90.5%), scoliosis (73.5%), feet deformities (58.8%), urinary dysfunction

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(42.8%), cardiomyopathy (40.3%) followed by decrease visual acuity (36.8%) [2].

Indelicato et al. reported in their study that, 90.7% of cases of Friedreich's ataxia presented with gait or coordination disturbance. Non neurological features or late adulthood presentation still leads to significant delay in the diagnosis of Friedreich's ataxia [3].

This goes with this particular presented case since, the presenting feature was no neurological; low back pain and the presentation was very late adulthood.

CONCLUSION

Chronic low back pain not responding to the usual treatment, particularly associated with balance and gait disturbance, could be, although rarely, a presenting feature of Friedreich's ataxia and full clinical and electrophysiological examination should be conducted to prove or disprove such diagnosis.

REFERENCES

1. Parkinson MH, Boesch S, Nachbauer W, Mariotti C, Giunti P. Clinical features of Friedreich's ataxia: classical and atypical phenotypes. J Neurochem. 2013;412(S1):103-117.
2. Reetz K, Dogan I, Hohsenfeld C, Didszun C, Guinti P et al. Non-ataxia symptoms in Friedreich's ataxia. Report from the registry of the European Friedreich's ataxia consortium for translational studies (EFACTS). Neurology 20184;91(10): e917-e930.
3. Indelicanto E, Nachbauer W, Eigenther A, Amprosi M, et al. Onset features and time to diagnosis in Friedreich's ataxia. Orphanet J Rare Dis. 2020;3;15(1): 198.