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Usefulness of Muscle MRI in the Diagnosis of Axial Myopathy: Two Case Reports

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Introduction

Myopathies are traditionally categorized by limb or cranial nerve involvement, but with the increased use of magnetic resonance imaging (MRI), many myopathies affect the axial musculature. Axial myopathy refers to diseases with selective involvement of axial muscles. Degeneration of paraspinal muscles in degenerative spinal diseases is often mistaken for neurogenic degeneration. Since creatine kinase (CK) and lactate dehydrogenase (LDH) levels are normal or mildly elevated, axial myopathy is often overlooked. This study presents two cases of axial myopathy to raise awareness.

Two Case Reports

Case 1 (F/56)

- Symptoms: 10-year history of lower back pain and progressive leg weakness
- Physical examination: proximal muscle weakness (MRC grade 3) in both limbs, along with bladder and bowel incontinence, waddling gait with knee flexion and a lean-back posture
- Lumbar MRI: severe fatty infiltration of paravertebral muscles (Figure 1)
- Electrodiagnostic findings: indicated bilateral L5-S1 radiculopathy
- Lab tests: CK 214 U/L (normal range: 21-215 U/L) and LDH 384 IU/L (normal range: 225-455 IU/L)
- Muscle MRI: diffuse fatty atrophy of paravertebral muscles, sparing the proximal multifidus muscles (Figure 3)
- Next-Generation Sequencing (NGS): heterozygosity for POMT1 and CHST14 (autosomal recessive) and AARS1 and NIPA1 (variants of unknown significance).
- Based on these findings, the patient was diagnosed with axial myopathy.

Case 2 (M/61)

- Symptoms: 10-year history of lower back pain and progressive leg weakness, diagnosed with spinal stenosis, underwent L4-L5-S1 posterior lumbar interbody fusion surgery.
- Physical examination: muscle weakness (MRC grade 4) in both lower limbs
- Lumbar MRI: fatty infiltration of paravertebral muscles (Figure 2)
- Electrodiagnostic findings: bilateral L5 radiculopathy.
- Lab tests: normal CK (65 U/L) and LDH (384 IU/L)
- Muscle MRI: fatty atrophy of multifidus (Figure 3)
- One month after surgery, his symptoms improved, and he is under observation for possible idiopathic inflammatory myopathy. If symptoms worsen, muscle biopsy and genetic testing will be considered.

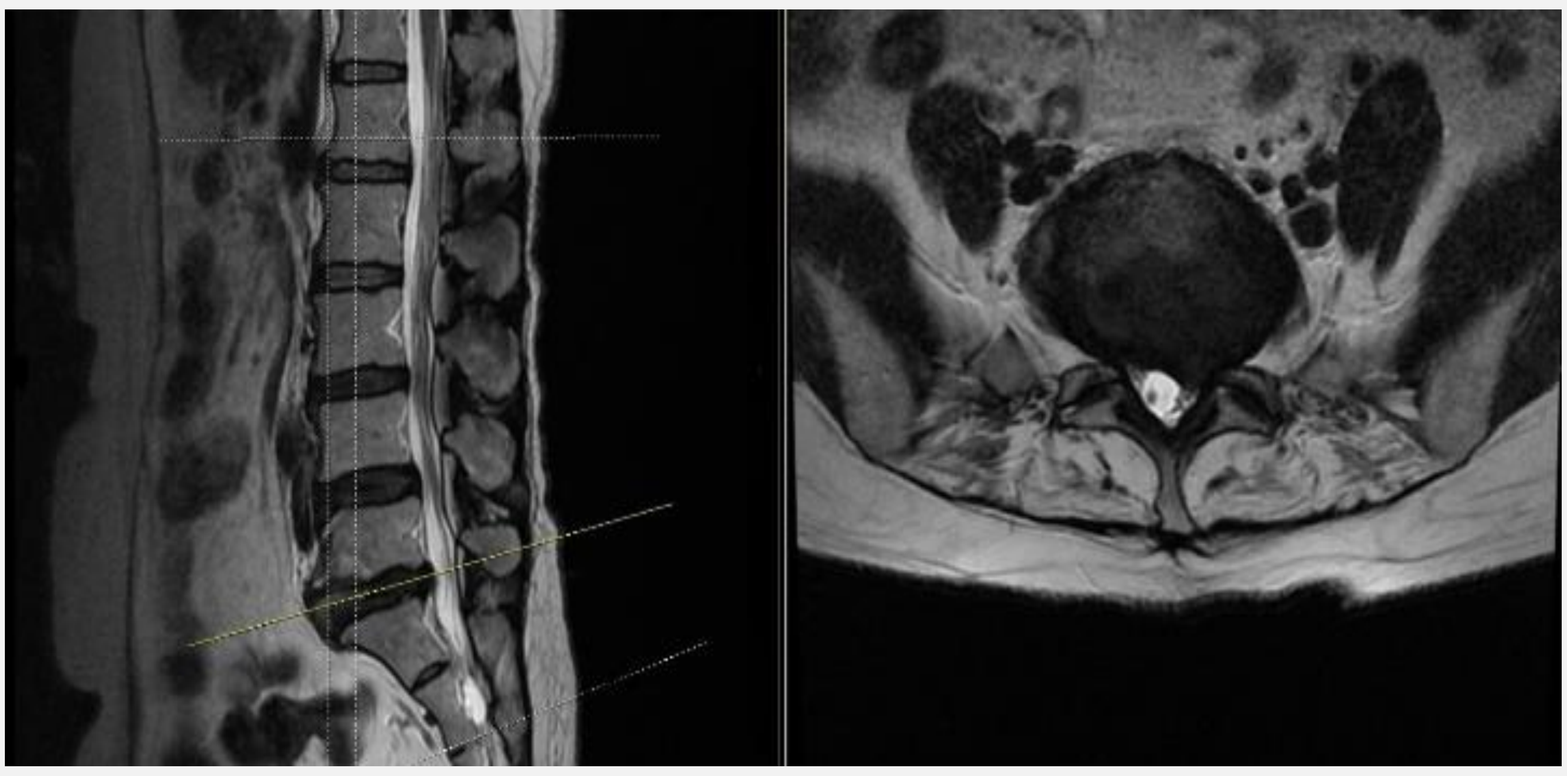

Figure 1. Lumbar MRI of Case 1 patient

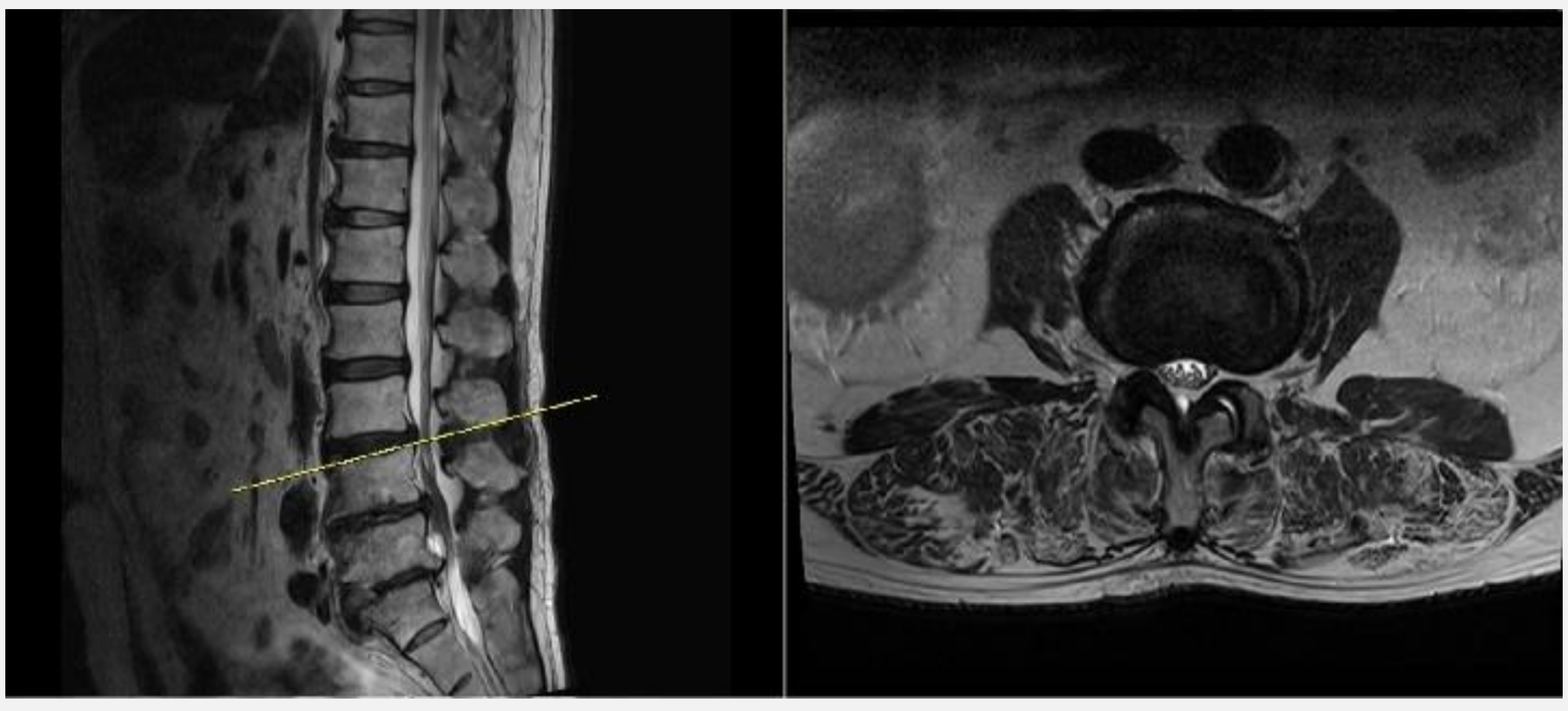

Figure 2. lumbar MRI of Case 2 patient

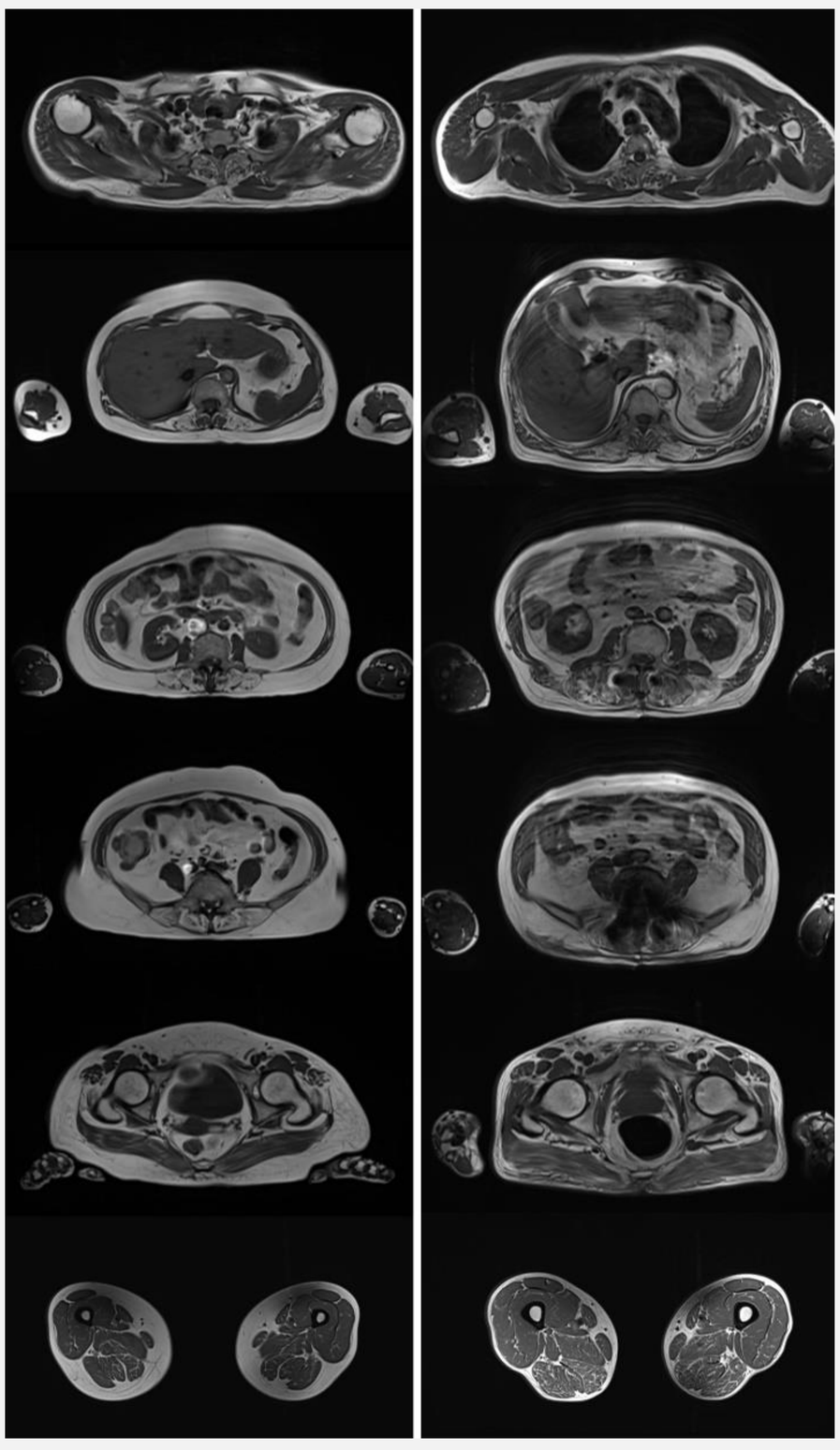

Figure 3. The T1 whole-body muscle MRI in the figure shows the T3, T10, L3, L5, femoral neck, mid-femur levels. Diffuse fatty atrophy was observed in the paravertebral muscles

Conclusion

Axial myopathy encompasses a group of diseases requiring further research into their distribution, diagnostic significance, clinical outcomes, and management. Studies suggest that electromyography, CK levels, muscle histology, and genetic testing aid diagnosis, with muscle MRI being helpful. Clinical attention should focus on paraspinal muscle involvement, particularly in patients with degenerative spinal diseases.