

When Occam's Razor Fails: A McArdle Disease Diagnosis Following Idiopathic Transverse Myelitis

Wendy Chen, Steven R. Dunham, Lydia Sharp – Department of Neurology, Baylor College of Medicine, Houston, Texas

Background

Idiopathic transverse myelitis¹ is a rare inflammatory spinal cord disorder that can present with weakness, numbness, or autonomic dysfunction.

McArdle disease², an autosomal recessive disorder caused by muscle phosphorylase deficiency, results in lifelong exercise intolerance, exertional rhabdomyolysis, muscle contractures, and a second-wind phenomenon.

Transverse Myelitis

A 44-year-old woman contracted gastroenteritis and developed ascending numbness a week later.

Labs including serum and CSF studies were unremarkable.

MRI brain: unremarkable.

MRI cervical spine: nonenhancing C5-C6 lesion.

MRI thoracic spine: mildly enhancing T7 lesion.

She was diagnosed with idiopathic transverse myelitis and received a course of steroids.

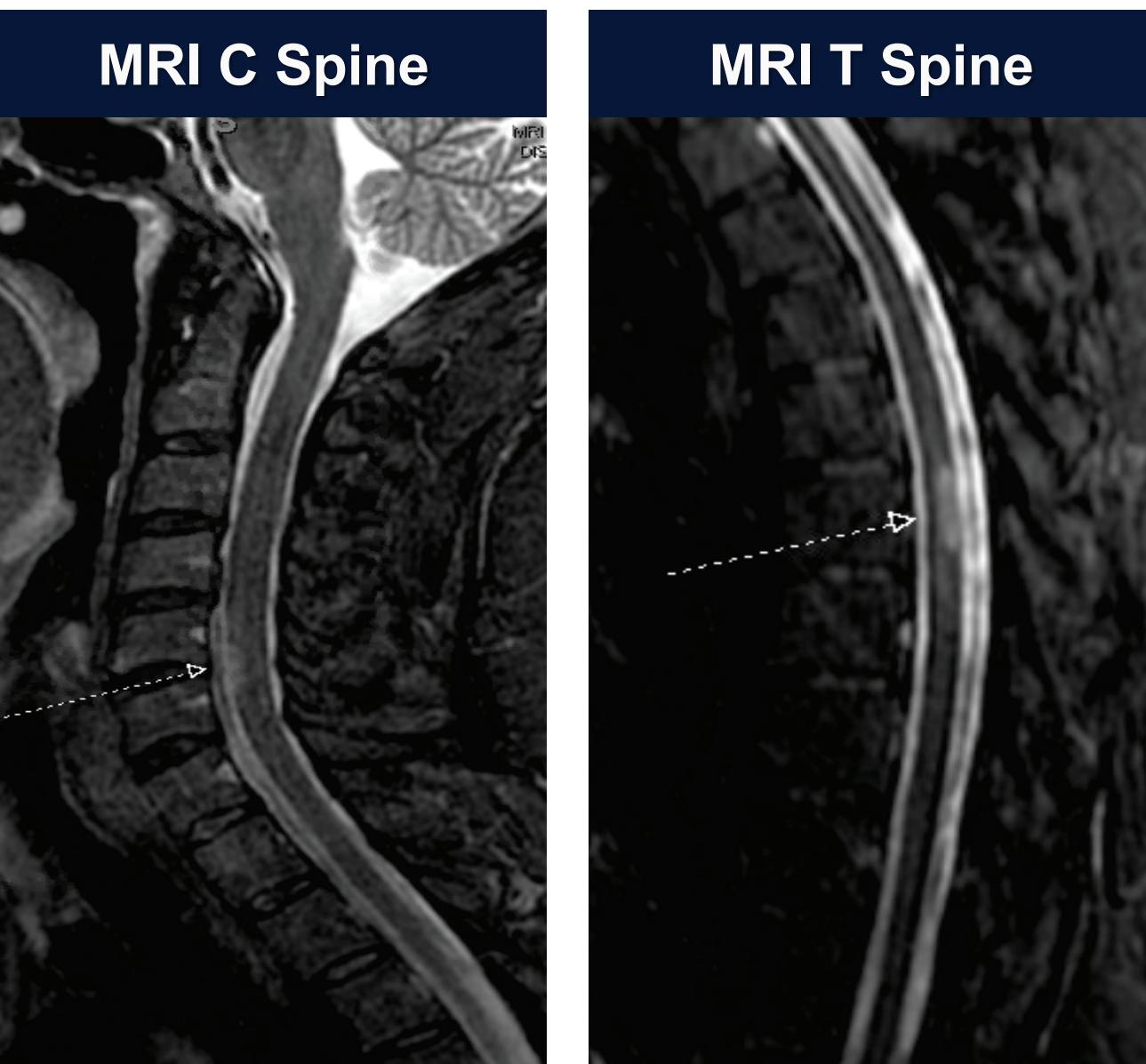
Patient was not started on DMT given that she did not meet diagnostic criteria for multiple sclerosis or appear to have relapsing disease.

Download poster

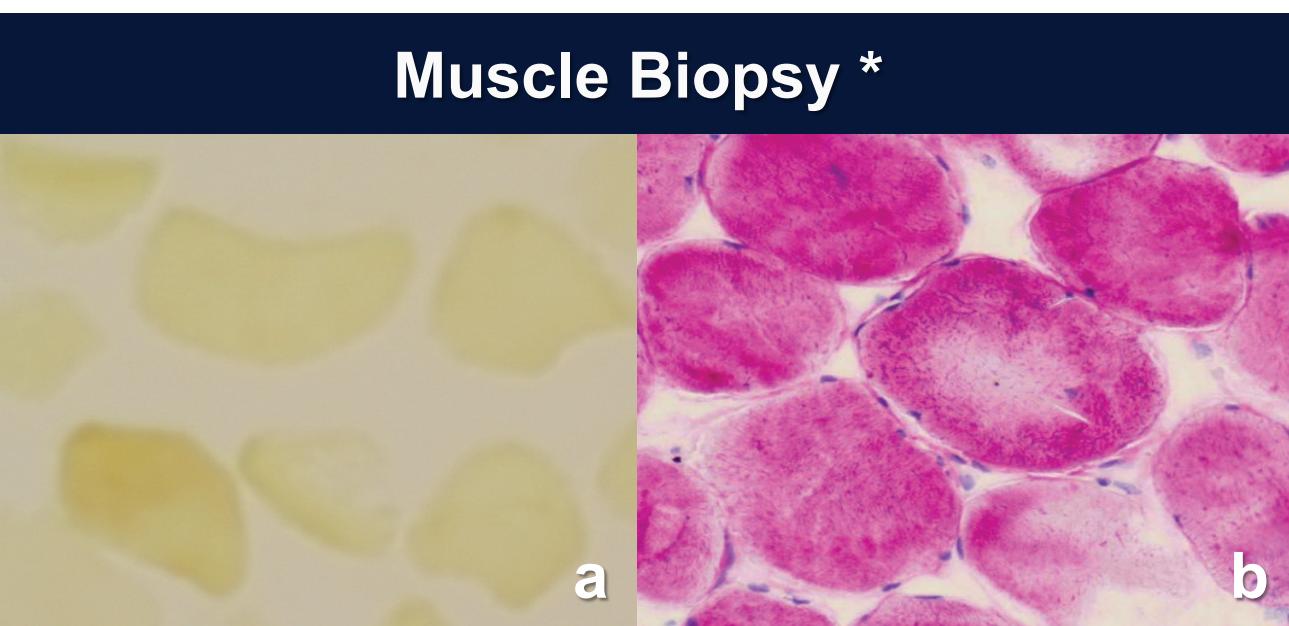


Workup

Serum Labs	Results
ANA	Initially positive, then negative
RF	Negative
Anti-cardiolipin	Negative
Anti-dsDNA	Negative
Anti-Smith	Negative
Anti-RNP	Initially positive, then negative
C3 / C4	Negative
ANCA	Negative
SSA / SSB	Negative
Thyroid Panel	Negative
B12	879
ESR / CRP	10 / 0.8
NMO IgG	Negative



CSF Labs	Results
RBC	2
WBC	3
Protein	41
Glucose	52
Gram Stain / Culture	Negative
VDRL	Negative



[a] Absent myophosphorylase stain
[b] PAS stain showing increased glycogen stores

* Courtesy of Dr. Kent Heck

Conclusions

- This is the first report of a patient with transverse myelitis and McArdle disease.
- Shang et al.³ reported a case of a woman diagnosed with NMOSD after rhabdomyolysis.
- Fatigue, weakness, and pain are common issues in both TM and McArdle disease.
- The two pathologies appeared to occur independently, but the truncal numbness from transverse myelitis might have masked the myopathic pain and contributed to later hospitalizations for rhabdomyolysis.
- Patient's spasticity related to TM might also have increased metabolic demand in skeletal muscle, lowering the threshold for rhabdomyolysis.

McArdle Disease

Several months following the episode of transverse myelitis, patient was hospitalized twice for rhabdomyolysis after developing severe back pain following exertion.

CK was elevated up to 40,000.

Muscle biopsy during the second admission showed absent myophosphorylase and increased glycogen stores, consistent with McArdle disease.

Upon further questioning, patient disclosed lifelong exercise intolerance, dark urine after exercise, contractures, and improved exercise capacity after rest.

She believed truncal numbness related to transverse myelitis might have concealed the warning signs of impending muscle contracture.

Occam's Razor: The simplest explanation is usually correct.

Hickam's Dictum: Patients can have as many diseases as they well please!

References

- de Seze J, Lanctin C, Lebrun C, Malikova I, Papeix C, Wiertlewski S, Pelletier J, Gout O, Clerc C, Moreau C, Defer G, Edan G, Dubas F, Vermersch P. Idiopathic acute transverse myelitis: application of the recent diagnostic criteria. *Neurology*. 2005;65(12):1950-3.
- Pallo PAO, Silva AMSd, Zanoteli E, Shinjo S. McArdle's disease: an underestimated or underdiagnosed myopathy in rheumatologic practice? Cases series and literature review. *MedicalExpress (São Paulo, online)*. 2018;5:mo18008.
- Shang K, Qin C, Bu BT, Tian DS. Aquaporin-4 antibody positive neuromyelitis optica spectrum disorder subsequent to rhabdomyolysis: a case report and literature review. *Int J Neurosci*. 2019;1-3.