Conus Medullaris Infarction in a Patient With Familial Mediterranean Fever: A Case Report

Zoe Robinow, BS1; Kathleen Barnett, BS2; John Geraghty, MD1,2

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ABSTRACT

Background: Spontaneous spinal cord infarctions are rare, especially in the conus medullaris (CM). They are a particularly uncommon presentation in patients with familial Mediterranean fever (FMF).

Case Description: Our patient is a 50-year-old man with FMF, controlled with colchicine for 20 years, who presented to the emergency department when he developed the inability to ambulate without assistance. He also had bowel and bladder incontinence after experiencing burning in his thighs, scrotum, and penis that radiated down his legs. A magnetic resonance imaging scan with and without gadolinium showed T2 hyperintensity changes in the CM and L2 vertebral body, with enhancement of the CM and cauda equina. The patient received high-dose steroids while hospitalized without clinical benefit. He noted improved strength over the past several months, particularly in his left leg, but has persistent sensory disturbances in his buttocks, scrotum, and plantar surfaces. He continues to experience bowel and bladder incontinence.

Conclusion: Although CM infarction is rare in patients with FMF, it should be considered in the differential diagnosis when there is a high index of suspicion. The presence of vertebral body infarction with T2 changes on magnetic resonance imaging will indicate similar pathology in the CM.

INTRODUCTION

Spontaneous spinal cord infarctions are infrequent, especially in the conus medullaris (CM) region. It is estimated that 1.6 to 7.2 spinal cord infarctions occur per 100,000 individuals yearly, with a slightly higher occurrence in females than males.1 Affected patients can develop lasting sequelae. CM infarction is a particularly rare presentation in patients with familial Mediterranean fever (FMF).

CASE NARRATIVE

Our patient is a 50-year-old mild to moderately obese male engineer with FMF, controlled with colchicine for 20 years. He was in his usual state of health until he noted exhaustion and burning in his thighs, scrotum, and penis that radiated down his legs. A magnetic resonance imaging scan with and without gadolinium showed T2 hyperintensity changes in the CM and L2 vertebral body, with enhancement of the CM and cauda equina. The patient received high-dose steroids while hospitalized without clinical benefit. He noted improved strength over the past several months, particularly in his left leg, but has persistent sensory disturbances in his buttocks, scrotum, and plantar surfaces. He continues to experience bowel and bladder incontinence.

Neurologic examination revealed a muscle strength of 4+/5 in the hip extensors, 4/5 in the right tibialis anterior, 5/5 in the left tibialis anterior, 4/5 in the bilateral extensor hallucis longus, 3/5 in the right plantar flexors, and 4/5 in the left plantar flexors. Otherwise, strength was 5/5 with normal tone. Reflexes were 2+ in the arms and knees, with absent ankle reflexes and flexor plantar responses bilaterally. There was decreased pinprick sensation in the left S2–S5 region, but sensation was intact on the right. Vibration sense was impaired in the great toes bilaterally.

The initial differential diagnosis included acute spinal cord infarction, transverse myelitis, and Guillain–Barre syndrome (GBS). Cerebrospinal fluid analysis performed in the emergency department showed a traumatic tap with a protein level of 50, a glucose level of 75, and 4 white blood cells. Serologies were negative for infectious etiology or heavy metal toxicity. Inflammatory markers, including erythrocyte sedimentation rate, C-reactive protein, and antinuclear antibody, and a hypercoagulable workup were within normal limits except for a mildly elevated protein C level. An electrocardiogram showed normal sinus rhythm with left atrial enlargement and evidence of old inferior and anteroseptal myocardial infarctions.

A noncontrast magnetic resonance imaging (MRI) scan of the lumbar spine was unremarkable. Follow-up MRI with and without gadolinium showed T2 hyperintensity changes in the CM and L2 vertebral body, with enhancement of the cauda equina and CM (Figures 1 and 2). These findings indicated an acute spinal cord infarction in the CM.

The patient received high-dose steroids while hospitalized without significant clinical benefit. His strength has slowly improved over the past several months, especially in his left leg. He has persistent sensory impairment in his buttocks, scrotum, and plantar surfaces of both feet. He takes pregabalin 100 mg 3 times daily for this, which provides

Abbreviations: CE, cauda equina; CM, conus medullaris; FMF, Familial Mediterranean Fever; GBS, Guillain-Barre syndrome; MRI, magnetic resonance imaging; WBC, white blood cell

Keywords: case report, conus medullaris, familial Mediterranean fever, spinal cord infarction

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modest benefit. However, he continues to experience bowel and bladder incontinence (Table 1).

**DISCUSSION**

FMF is an autosomal recessive autoinflammatory disease that presents with recurrent, sporadic attacks of fever and serosal inflammation, including peritonitis, pleuritis, or acute synovitis. Episodes can be triggered by emotional stress, fatigue, surgery, menstruation, exercise, or cold temperature and typically last for 1 to 3 days before spontaneously resolving. A variety of mutations of the *MEFV* gene on the short arm of chromosome 16 are implicated in FMF. The gene product, pyrin, is suspected to be involved in the regulation of inflammatory responses.

Neurologic complications are rarely seen in FMF, and few studies have examined the occurrence of stroke in affected patients. Of 18 patients with FMF and central nervous system involvement, Kalyoncu et al reported 7

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Table 1. Timeline of the patient’s clinical course

<table>
<thead>
<tr>
<th>Date</th>
<th>Summary</th>
<th>Diagnostic testing</th>
<th>Interventions</th>
</tr>
</thead>
<tbody>
<tr>
<td>April 12, 2020</td>
<td>Patient notes exhaustion and burning in legs, which later resolved.</td>
<td>n/a</td>
<td>n/a</td>
</tr>
<tr>
<td>April 13, 2020</td>
<td>Patient develops burning, weakness, and incontinence. Goes to the ED and is admitted.</td>
<td>• Traumatic lumbar puncture but protein level of 50, glucose level of 75, and 4 WBCs • Slightly elevated protein C; otherwise hypercoagulable, inflammatory, infectious, and heavy metal serologies unremarkable • ECG shows normal sinus rhythm, left atrial enlargement, old inferior MI, and old anteroseptal MI • CT negative for abdominal aortic aneurysm • Initial noncontrast MRI negative • Follow-up MRI with and without Gd demonstrates infarction in the conus medullaris</td>
<td>• High-dose steroids given in hospital; no significant clinical benefit • Prescribed pregabalin (Lyrica)</td>
</tr>
<tr>
<td>July 21, 2020</td>
<td>At follow-up visit, the patient continues to experience sensory deficits and bowel and bladder incontinence. Strength is improved.</td>
<td>n/a</td>
<td>• Continues to take pregabalin for modest benefit</td>
</tr>
</tbody>
</table>

CT, computed tomography; ECG = electrocardiogram; ED = emergency department; Gd = gadolinium; MI = myocardial infarction; MRI = magnetic resonance imaging; n/a = not applicable.
with cerebrovascular disease. Topcuoglu et al. investigated 17 acute ischemic strokes in 14 patients with FMF. Additionally, Luger et al. and Aghdashi et al. discussed 2 young men with FMF who experienced a brainstem infarction and a right periventricular infarction, respectively. In the case of a 13-year-old girl with FMF who suffered a right medullary stroke, Aoun et al. suggested she may have inherited and acquired prothrombotic risk factors. None of these studies reported on spinal cord infarction in patients with FMF.

The spinal cord is resistant to ischemia because of its strong vascularization. The anterior two-thirds is supplied by the single anterior spinal artery and supplemented with 6-8 radicular arteries from the aorta. The largest radicular artery is the artery of Adamkiewicz, which joins the anterior spinal artery primarily between T8 and L2. The dual posterior spinal arteries supply the posterior one-third of the spinal cord. This anatomy explains why spinal cord infarction most commonly affects the midthoracic area and why CM infarction is among the rarest infarction sites. Among the most common causes of spinal cord infarction in adults are atherosclerosis, thoracoabdominal aneurysms, emboli, hypotension, arteriovenous malformations, and sickle cell disease.

In our patient, the differential diagnosis included acute spinal cord infarction, transverse myelitis, and GBS. Transverse myelitis is associated with inflammatory diseases and infection. Similarly, GBS is associated with preceding diarrheal illnesses caused by organisms like *Campylobacter*. All of these pathologies can present with pain, sensory and motor changes, and bowel and bladder dysfunction. An MRI provides the most diagnostic information. Acute spinal cord infarction may show signal change in the vertebral body on T2 images, as seen in this patient. In GBS, there is typically contrast enhancement in the CM and cauda equina nerve roots. MRI findings for transverse myelitis are relatively nonspecific, including poorly delineated hyperintense signal on T2.

**CONCLUSION**

In summary, it is important to consider spinal cord infarction, including in the CM, in the differential diagnosis when a patient presents with sudden onset of symptoms, suggesting an acute ischemic event. The presence of vertebral body infarction on MRI is diagnostic of a spinal cord infarction.

**Disclosure Statement**

The author(s) have no conflicts of interest to disclose.

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None.

**Author Contributions**

Zoe Robinow, BS, and Kathleen Barnett, BS, assisted in the project design and wrote the manuscript. John Geraghty, MD, supervised the project and helped with data collection.

**Supplemental Material**

*Supplemental Material is available at: www.thepermanentejournal.org/files/2021/21.029supp.pdf*

**References**