A Mysterious Squeezing Pain

Jamie Bowen MD, Mihaela Tiru MD, Benjamin Barnes MD

Baystate Health | University of Massachusetts Medical School UMASS

BACKGROUND

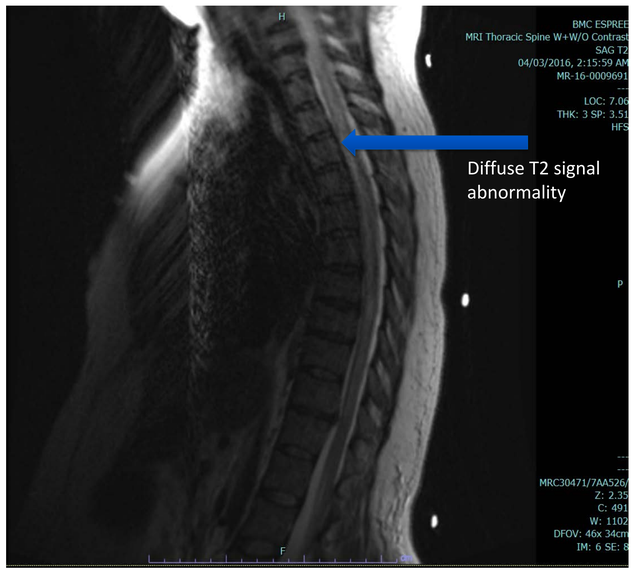
- Neuromyelitis optica (NMO) is an idiopathic, inflammatory demyelination disease of the central nervous system that causes severe optic neuritis and myelitis attacks
- Acute optic neuritis may be the first sign of multiple sclerosis (MS), but also of neuromyelitis optica
- Early diagnosis and distinction between MS and NMO is critical to cfacilitate correct treatment and prevention of future episodes
- There is a known association between Aquaporin 4 seropositivity and neuromyelitis optica[1]

CASE

- 46-yo female with a history of unilateral optic neuritis over 20 years prior without residual visual deficit presented with one week of increasing neurological symptoms
- She presented at outside hospital 1 week prior to this admission with epigastric pain and band-like tightness around her chest, was diagnosed with biliary colic, and had laparoscopic cholecystectomy
- Postoperatively, she developed paresthesias from mid-chest to bilateral legs and feet, urinary retention, and increasing impairment of bilateral lower extremity coordination.
- She was initially treated with empiric antiviral and antibacterial agents as well as high-dose IV steroids
- An extended CSF infectious panel by PCR was negative and all antimicrobial medications were discontinued
- T-Spine MRI showed abnormal T2 signal in the thoracic spine with small T10 disc protrusion and diffuse thoracic myelopathy which suggested a diagnosis of neuromyelitis optica[2].

RESULTS

Cerebrospinal fluid		Reference
WBC	124 cells/mm^3	*
Neutrophils	66%	*
Lymphocytes	30%	*
Protein	167 (H) mg/dL	15-30 mg/dL
Glucose	71 mg/dL	40-80 mg/dL
Myelin basic protein	>112.5 (H) ng/mL	0-1.2 ng/mL
Oligoclonal bands	8	*
IgG	185 (H) mg/L	10-30 mg/L
M/E PCR panel*	Negative	Negative



Thoracic MRI T2 Image

CASE CONTINUED

- She had minimal improvement with steroids so she underwent several rounds of plasma exchange for a presumed diagnosis of NMO. She had minor improvement during her hospitalization.
- Her serum aquaporin-4 antibody was positive confirming the diagnosis of neuromyelitis optica.
- 3 years later the patient has near complete motor function and moderately improved sensory function.
- She remains on maintenance regimen of rituximab every 6 months with intermittent steroids for disease flares

KEY POINTS

- It is important to have high clinical suspicion of autoimmune neurological diseases in patients with even a remote history of optic neuritis. MRI can be used to quickly differentiate between NMO and MS, leading to expedited treatment of acute episodes.
- It is reasonable to test all patients with a history of optic neuritis for AQ4-Ab as the diagnosis of NMO leads to different maintenance and suppressive therapy than multiple sclerosis. The consequences of an acute flare can lead to significant morbidity, including permanent neurological disability.
- Aquaporin-4 antibody seropositivity in patients with a history of uncomplicated monocular optic neuritis has been shown to correlate with increased severity of optic neuritis episodes and future development of NMO. There was no increased risk of development of multiple sclerosis.[1]

References

1. Jarius S, Frederikson J, Waters P, Paul F, Akman-Demir G, Marignier R, Franciotta D, Ruprecht K, Kuenz B, Rommer P, Kristoferitsch W. Frequency and prognostic impact of antibodies to aquaporin-4 in patients with optic neuritis. Journal of the neurological sciences. 2010 Nov 15;298(1-2):158-62.
2. Lalan S, Khan M, Schlakman B, Penman A, Gatlin J, Herndon R. Differentiation of neuromyelitis optica from multiple sclerosis on spinal magnetic resonance imaging. Int J MS Care. 2012;14(4):209–214. doi:10.7224/1537-2073-14.4.209
3. Kim SM, Kim SJ, Lee HJ, Kuroda H, Palace J, Fujihara K. Differential diagnosis of neuromyelitis optica spectrum disorders. Ther Adv Neurol Disord. 2017;10(7):265–289. doi:10.1177/1756285617709723

The Case of the Stiff Man

Ritika Walia, MD and Cortney Haynes, MD

Introduction

- Stiff Person Syndrome (SPS) is a rare neurological entity characterized by progressive muscle rigidity and painful spasms
- Epidemiology: SPS has a prevalence of 1 case per 1 million individuals, with a 2:1 female:male predominance
- Here, we present a male patient in whom symptoms began in a single extremity and rapidly generalized

Case Presentation

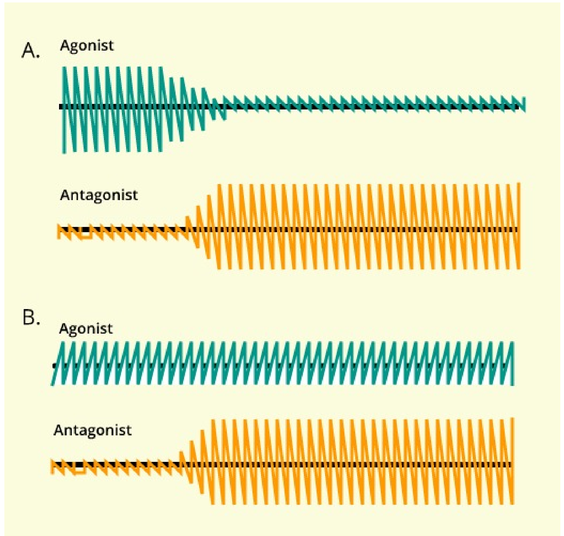
HPI
- 59 yo M presented to his physician complaining of right ankle pain and stiffness
- Over the next 8 months, symptoms spread to involve both legs and the lower back
- Associated symptoms included dyspnea and unintentional weight loss; ROS otherwise negative

PMH
- Vitiligo, benign prostatic hypertrophy, hypertension
- Up to date on all age-appropriate cancer screenings

Exam
- Marked stiffness in the ankle, thighs, and lower back joints
- Severe pain with movement
- Diminished strength in the bilateral lower extremities
- Positive Hoffman's sign on the left foot

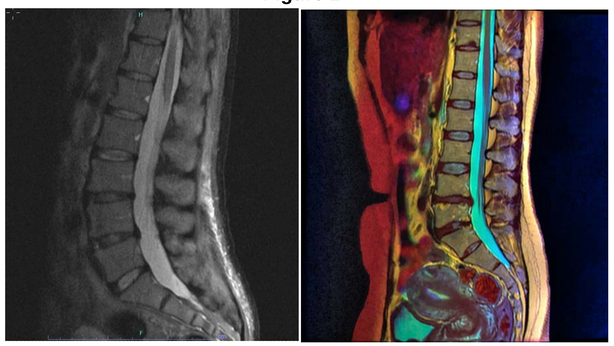
Dx and Treatment
- MRI: mild degenerative changes
- GAD65 antibodies present in high titers
- Started on high dose diazepam and referred to a movement disorders specialist

Figure 1



A. Schematic displaying normal inhibition of muscle contraction when the antagonist muscle fires, versus B. simultaneous, sustained, and involuntary contraction of agonist muscle despite voluntary activation of antagonist muscle.

Figure 2



Lumbar spine MRI images, showing the patient's normal lumbar lordosis (left), and loss of lumbar lordosis (right) which can occur as a complication of frequent muscle spasms.

Discussion

- SPS is characterized by excessive firing of motor neurons and simultaneous sustained contraction of agonist and antagonist muscles[1]
- The classic subtype manifests in the axial and proximal limb muscles, whereas distal-limb predominant symptoms are seen in the partial subtype. Less than 2% of SPS cases are paraneoplastic[2]
- Antibodies against glutamic acid decarboxylase (GAD65) are found in 80% of patients with classic SPS; lead to disinhibition of the GABA pathway[3]
- Typically treated with high dose benzodiazepines and IvIg[2]
- Depression, anxiety, and agoraphobia are common[2]

Conclusions

- While rare, SPS is important to consider in the differential for muscle stiffness and frequent spasms
- This patient displayed characteristics of both classic and partial SPS, along with history concerning for the paraneoplastic variant
- The psychological manifestations of rapid functional decline reflect a significant component of the burden of this disorder

References

1. Rakocevic, G., & Floeter, M. K. (2012). Autoimmune stiff person syndrome and related myelopathies: Understanding of electrophysiological and immunological processes: Stiff Person Syndrome. *Muscle & Nerve*, *45*(5), 623–634. https://doi.org/10.1002/mus.23234
2. Ciccotto, G., Blaya, M., & Kelley, R. E. (2013). Stiff person syndrome. *Neurologic Clinics*, *31*(1), 319–328. https://doi.org/10.1016/j.ncl.2012.09.005
3. Holmøy, T., & Geis, C. (2011). The immunological basis for treatment of stiff person syndrome. *Journal of Neuroimmunology*, *231*(1–2), 55–60. https://doi.org/10.1016/j.jneuroim.2010.09.014
4. Dilmen, N. (2016). *English: Magnetic resonance imaging of the spine*. Retrieved from https://commons.wikimedia.org/w/index.php?title=File:Lumbar_MRI_T1FSE_T2frFSE_STIR_08.jpg&oldid=299147084.

TOO SMALL FOR STONES: AN UNCOMMON ETIOLOGY FOR A COMMON PROBLEM

Nicholas Karr MD[1,2] , Deirdre Lewis MD[1,2], Jason Phillippi MD[1,2]
[1] Department of Medicine, [2] Department of Pediatrics,
University of Massachusetts Medical School-Baystate, Springfield MA

INTRODUCTION

1. Gallstones is a common cause of abdominal pain in adults but uncommonly seen in pediatrics
2. There are specific conditions that can precipitate gallstone formation in children
3. Management depends on underlying pathology

CASE PRESENTATION

- 4-year-old girl with a one-year history of constipation and failure to thrive followed by gastroenterology, presented to the ED with sudden onset abdominal pain.
- Initial workup ruled out intussusception, ovarian torsion and appendicitis and patient was discharged.
- Returned three days later with unresolved worsening sharp abdominal pain and new onset non-bloody non-bilious vomiting, scleral icterus and pale stools.
- Review of systems was negative for fevers, upper respiratory infection symptoms, diarrhea, myalgias, cough and recent weight loss.
- Stable vital signs without fever. Physical exam revealed an ill appearing child who was uncomfortable. Pertinent findings included a soft non-distended abdomen, active bowel sounds, and generalized abdominal pain on superficial and deep palpation without rebound.
- Right upper quadrant ultrasound revealed diffuse fatty infiltration of the liver, common bile duct at 6mm with trace nonspecific perihepatic ascites.
- Lab values were significant for ALT/AST of 201/76, lipase of 34, alkaline phosphatase of 309, total bilirubin of 3.8 with direct bilirubin of 2.7, and GGTP of 121. PT, CBC and electrolytes were all within normal range.
- Patient admitted for further management.
- MRCP showed dilated common bile duct at 11mm, biliary sludge and two stones.
- Then underwent stone removal via ERCP without sphincterotomy. After removal of biliary sludge and 2 stones, her clinical symptoms improved, she was pain free with no jaundice or acholic stools and she was discharged home with GI follow up.

RADIOGRAPHIC IMAGING

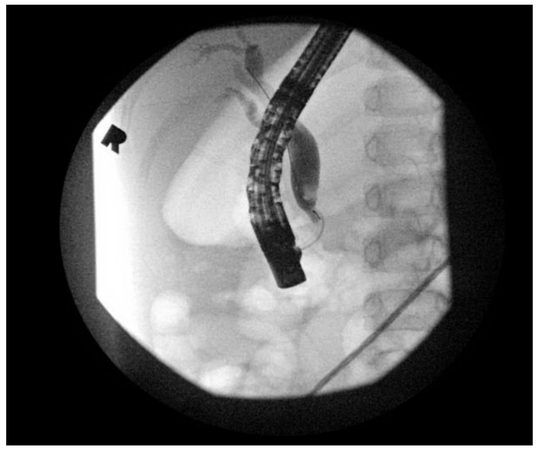


Figure 1: ERCP showing common bile duct dilation

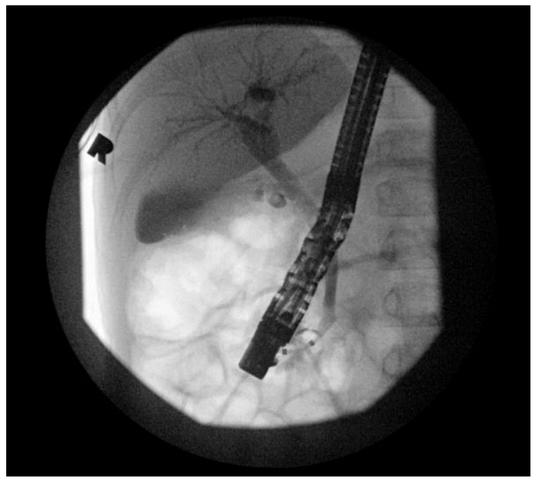


Figure 2: ERCP after papillotomy showing reduced common bile duct dilation

DISCUSSION

-Abdominal pain one of most common complaints in pediatrics, however choledocholithiasis is a relatively uncommon cause.
-Incidence of gallstones is unknown in pediatric patients, however it appears to be increasing
-Increase likely multi-factorial including increased short gut survivorship, increased obesity, and improved diagnostic techniques.
-Many of the same risk factors have been found to be present in children as in adults for gallstones. Risk factors include obesity, type II diabetes, family history and female gender.
-Certain diseases increase risk including thalassemia, sickle cell, cirrhosis, Crohn's disease, and any cause of hyperbilirubinemia.
-In some patients such as in this case no risk factor is identified
-It is possible that gallstones formed as a result of gallbladder stasis associated with failure to thrive with biliary sludge present on ultrasound
-ERCP is not often used in pediatrics. Several studies have shown with appropriate training it is safe and efficacious for the management of multiple pancreatic and biliary disorders in neonates and children.

CONCLUSIONS

Gallstones are an uncommon cause of abdominal pain in pediatric patients. Risk factors for gallstone in children include family history, obesity, type 2 diabetes, female gender, and underlying disease. ERCP, though uncommon in pediatrics, may be successfully used to manage gallstones in children with an experienced operator.

REFERENCES

- Frybova B, Drabek J, Lochmannova J, et al. Cholelithiasis and choledocholithiasis in children; risk factors for development. PLoS One. 2018;13(5):e0196475. Published 2018 May 15. doi:10.1371/journal.pone.0196475
- Guralnick, S. Cholelithiasis and Cholecystitis. Pediatrics in Review. Sep 2009, 30 (9) 368-369; doi: 10.1542/pir.30-9-368
- Suh SG, Choi YS, Park KW, Lee SE. Pediatric cholecystectomy for symptomatic gallstones unrelated to hematologic disorder. Ann Hepatobiliary Pancreat Surg. 2016;20(4):187–190. doi:10.14701/ahbps.2016.20.4.187
- Usatin D, Fernandes M, Allen IE, Perito ER, Ostroff J, Heyman MB. Complications of Endoscopic Retrograde Cholangiopancreatography in Pediatric Patients; A Systematic Literature Review and Meta-Analysis. J Pediatr. 2016;179:160–165.e3. doi:10.1016/j.jpeds.2016.08.046