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TEACHING FILES (GRAND ROUNDS)

CONSTIPATION AS A PRESENTATION OF DUCHENNE MUSCULAR DYSTROPHY

Dhruv Gandhi, Ira Shah.

Department of Pediatric Gastroenterology and Hepatology, BJ Wadia Hospital for Children, Mumbai, India.

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chronic intestinal pseudo-obstruction, gastric dilatation, sigmoid volvulus, bowel dysmotility, genetic myopathies, whole exome sequencing.

Clinical Problem:
An 8-year-old boy presented in November 2024 with abdominal distention, intermittent vomiting, and an inability to spontaneously pass stools requiring multiple enemas for 1 month. He had progressive inability to get up from a squatting position starting at 3 years. He was evaluated at another centre for suspected muscular dystrophy and was found to have a creatine phosphokinase (CPK) of 17,000IU/L(normal: 10-120 IU/L). On presentation, his weight was 14 kg (<3rd percentile according to the Indian Academy of Pediatrics (IAP) charts) and his height was 105 cm (<3rd percentile according to the IAP charts). On examination, he had pallor, myopathic facies, baggy pant appearance, severe proximal muscle wasting, calf pseudohypertrophy, and Gower's sign was positive. There was upper abdominal fullness. Abdominal ultrasound showed grossly-loaded colon loops. Abdominal contrast-enhanced computerized tomography showed redundant recto-sigmoid colon dilated with fecal loading and mild circumferential thickening of the descending colon. CPK at our centre was 287 IU/L. Nerve conduction studies were suggestive of axonal motor neuropathy of bilateral upper and lower limbs. Electromyography was suggestive of a mixed myopathic-neuropathic pattern. Stool routine showed 80-100 pus cells/high power field and stool culture grew multidrug-resistant Escherichia coli sensitive to carbapenems. Stool Xpert MTB/Rif was negative and stool calprotectin was 5890 g/gm (normal: <50 g/gm). He was treated with intravenous meropenem. Ileocolonoscopy showed a distorted ileocaecal valve, active colitis with pseudopolyps, multiple deep ulcers in the caecum and the entire colon. Colon biopsy showed cryptitis but no crypt abscesses were present. He received oral mesalamine, azathioprine, prednisolone(2 mg/kg/day), and laxatives, to which he symptomatically responded, and was discharged on day-20. Whole exome sequencing revealed a likely pathogenic X-linked hemizygous deletion [c.(7098+1_7099-1)_(7309+1_7310-1)del] in the DMD gene at exons 49-50, suggestive of Duchenne muscular dystrophy (DMD). In view of the diagnosis of DMD, he was suspected to have chronic intestinal pseudo-obstruction (CIPO). Steroids were tapered, mesalamine dose was reduced, and azathioprine was stopped. He was started on oral rifaximin and probiotics.

What are the gastrointestinal manifestations in DMD and how to treat them?

Discussion:
DMD is an X-linked recessive disorder caused by mutations in the dystrophin gene located at Xp21.[1,2] Dystrophin has been demonstrated to be expressed in the smooth muscle cells of the gastrointestinal tract and postmortem studies have shown smooth muscle fibrosis and bowel wall atrophy in DMD patients.[2] Additionally, Lo Cascio et al.[1] demonstrated delayed gastric emptying time, colonic transit time and oro-caecal transit time in patients of DMD compared to normal individuals. Thus, dystrophin deficiency is the primary factor responsible for the gastrointestinal manifestations in DMD.[2] Visceral smooth muscle weakness can result in the following gastrointestinal manifestations in DMD: esophageal dysmotility and dysphagia, gastro-esophageal reflux, gastroparesis and gastric dilatation, subileus, CIPO, constipation, and volvulus.[1,2,3,4,5] Esophageal dysmotility can lead to dysphagia, loss of appetite and subsequent dehydration and malnutrition.[2] Delayed gastric emptying can result in gastroparesis and reflux, which in turn can predispose these patients to aspiration pneumonia, which may be fatal. Other risk factors such as steroid use, mechanical ventilation, immobility and scoliosis may also contribute to reflux in DMD patients.[2,3] Acute gastric dilatation warranting nasogastric decompression has also been reported in DMD patients.[2] Chronic constipation is a common manifestations of DMD with its prevalence estimated to be around 47%.[4] Several risk factors may contribute to this including steroid use, calcium supplementation, abdominal wall weakness, immobility, dyselectrolytemia, and mechanical ventilation.[2,3] Mechanical ventilation is particularly significant, as aerophagia can result in gastric and intestinal dilatation, which subsequently leads to abdominal distention and hinders adequate lung expansion. This, in turn, warrants the need for higher

Address for Correspondance: Dhruv Gandhi, 5B/13 Shyam Niwas, Breach Candy, Mumbai-400026, Maharashtra, India.
Email: dhruvgandhi2610@gmail.com

inspiratory pressures, worsening the aerophagia in a vicious cycle.[2] CIPO and sigmoid volvulus are potentially life-threatening complications seen in DMD patients. CIPO results from gut dysmotility and subsequent dilatation of an affected gut segment. Colonic dilatation in CIPO predisposes the patient to volvulus, which can lead to bowel obstruction, ischemia, and perforation. CIPO and recurrent volvulus can lead to reduced oral intake and contribute to the poor nutritional status of DMD patients.[5] While the management of gastrointestinal complications in DMD does not significantly differ from the management in patients without DMD, invasive measures and multimodality care may be required. Enemas and laxatives including bisacodyl, polyethylene glycol, and fibre supplements are routinely employed to treat constipation. Linaclotide, prucalopride and magnesium hydroxide may also be used. Prokinetics such as erythromycin, metoclopramide, and domperidone are useful in gastroparesis.[2] Ventilator settings should be optimised to reduce aerophagia and air removal should be done via a venting percutaneous endoscopic gastrostomy (PEG) tube placement or decompressive rectal tube placement. Nutritional modifications including diet optimisation, avoidance of beans, carbonated drinks and gum, food and fluid charting, enteric feeding via nasogastric tube or PEG tube, and total parenteral nutrition. Neostigmine is used in cases of acute intestinal pseudo-obstruction and rifaximin and metronidazole are used in case of small intestinal bowel obstruction.[2]

Compliance with ethical standards
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Conflict of Interest: None

References:

1. Lo Cascio CM, Goetze O, Latshang TD et al. Gastrointestinal Dysfunction in Patients with Duchenne Muscular Dystrophy. PLoS One. 2016 Oct 13;11(10):e0163779.
2. Blokhuis AM, Tytgat K, Groothuis JT et al. Severe gastrointestinal problems in Duchenne muscular dystrophy: A case series. Neuromuscul Disord. 2024 Jul;40:31-37.
3. Birnkrant DJ, Bushby K, Bann CM et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. Lancet Neurol. 2018 Mar;17(3):251-267.
4. Kraus D, Wong BL, Horn PS et al. Constipation in Duchenne Muscular Dystrophy: Prevalence, Diagnosis, and Treatment. J Pediatr. 2016 Apr;171:183-8.
5. Nart L, Desikan M, Emmanuel A et al. Life-threatening bowel complications in adults with Duchenne muscular dystrophy: a case series. Neuromuscul Disord. 2023 Nov;33(11):873-876.