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Transverse colonic volvulus in a 16-year-old female with congenital myotonic dystrophy: A case report

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ABSTRACT

Transverse colonic accounts for 2–4% of all forms of colonic volvulus and has its highest mortality rate (33) [1–3]. Only forty cases of pediatric transverse colonic volvulus have been reported in the literature to date [4]. We report on a 16-year-old female with congenital myotonic dystrophy who underwent operative repair for transverse colonic volvulus, the first reported case of these two entities in combination.

1. Background

Congenital myotonic dystrophy is a multisystem disorder, caused by progressive generational expansion of CTG trinucleotide repeats in the 3′ untranslated region of the Myotonic Dystrophy Protein Kinase (DMPK) gene located on chromosome 19q13.3, a locus that normally encodes for a serine threonine protein kinase involved in excitation-contraction coupling of muscle. As the number of CTG repeats increases, the symptoms associated with this disease, including the diagnosis-defining muscle wasting, weakness, and myotonia, also do [5]. Twenty-five percent of patients with myotonic dystrophy identify its gastrointestinal symptoms, which can manifest as dysfunction anywhere from the oropharynx to the anal sphincter, as the most disabling part of the disease. These symptoms include dysphagia, nausea, vomiting, bloating, diarrhea, and constipation [6–8]. Despite this fact, it is rare that surgical intervention of any kind is required for complications related to the gastrointestinal aspects of this disease. While several lines of evidence exist, the etiology of these gastrointestinal manifestations of myotonic dystrophy remain unclear.

2. Case report

A 16-year-old female with congenital myotonic dystrophy and chronic constipation presented to the emergency room complaining of severe, sharp, left lower quadrant, epigastric, and periumbilical, non-radiating abdominal pain for five days. The pain was associated with nausea, occasional vomiting, and decreased appetite, and was accompanied by two days of exacerbated constipation, refractory to treatment with oral hyoscamine and propylene glycol. Upon presentation to the emergency room, marked abdominal distention was noted on physical exam in addition to classic physical findings consistent with myotonic dystrophy (i.e. profound muscle wasting, muscle weakness, and myotonia). Since the abdominal x-ray (Fig. 1) showed a dilated colon loop in the right upper quadrant (bent inner tube/omega sign) and evidence of small bowel obstruction, nasogastric tube decompression, intravenous fluid resuscitation and pain control were initiated, with only mild relief of her symptoms. Computerized tomography of the abdomen (Fig. 2) demonstrated swirling of the mesenteric vasculature, dilation of the ascending and proximal transverse colon extending to the mid-transverse colon with tapering of the distal colon, consistent with transverse colonic volvulus. After urgent colonoscopic decompression was successful in detorsing the volvulus, the patient was taken to the operating room. Operative findings included malfixation of the cecum, right colon, and proximal transverse colon, with transverse and sigmoid colon redundancy noted in

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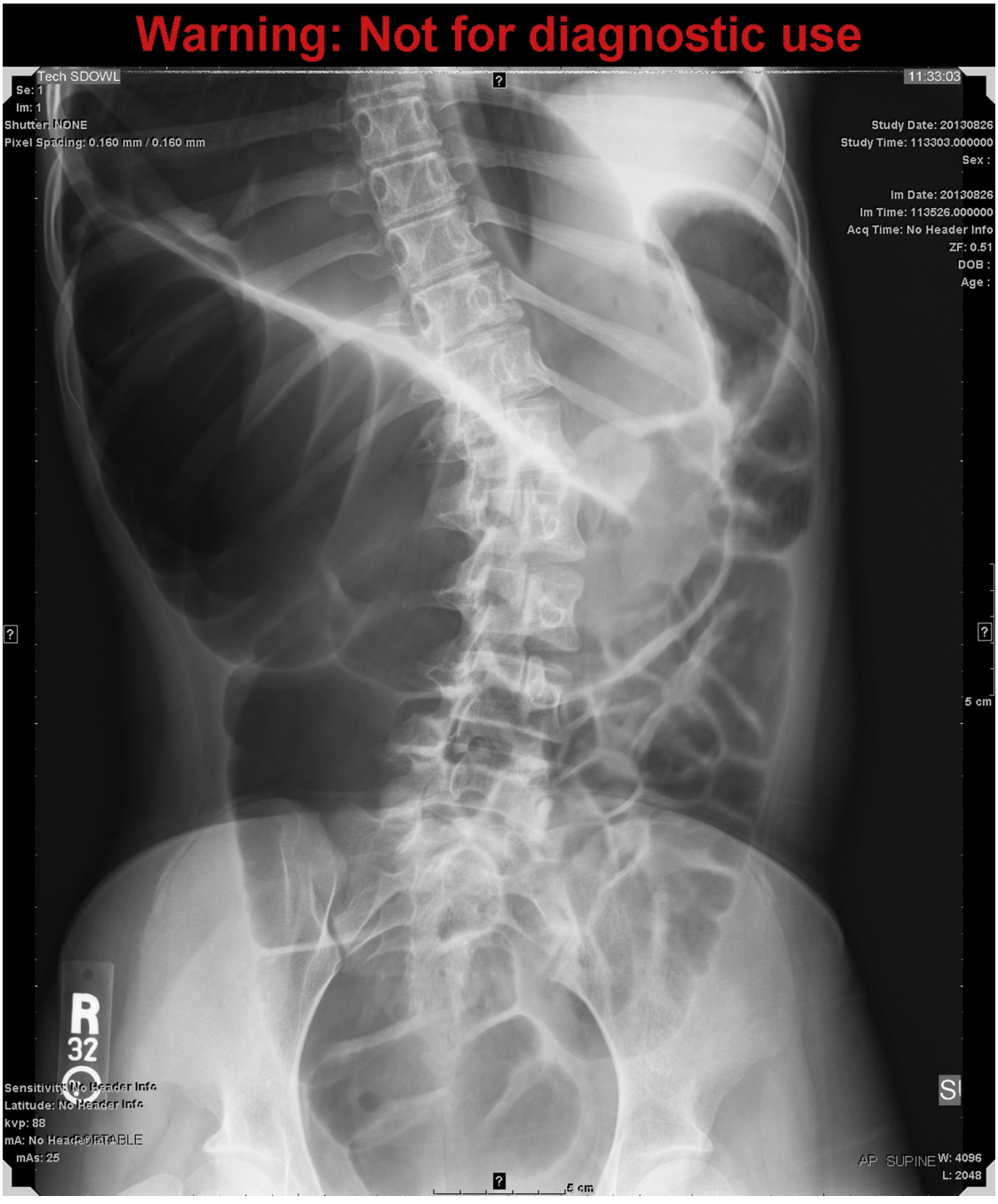

Fig. 1. Dilated colon loop in the right upper quadrant (bent inner tube/omega sign).

addition to attenuation of the descending colon. The patient underwent an extended right hemicolectomy with an ileo-distal transverse colon primary anastamosis.

3. Discussion

Similar to all other forms of colonic volvulus, transverse colonic volvulus, while extremely rare, is preceded by colonic redundancy and a narrow, parietally attached mesentery. However, this clinical entity is usually accompanied by congenital bands, distal obstructing lesions or pregnancy, none of which were present in this patient [9]. Consistent with most adult patients with any type of colonic volvulus, and with many patients with myotonic dystrophy, this patient had chronic constipation [6,9]. The cause of this problem in myotonic dystrophy is unknown but has been suggested to derive from disturbed neuropathic/channelopathic or neuroendocrine function in the gastrointestinal tract muscularis and/or somatic mosaicism/combined heterozygosity with other forms of muscular disease (e.g. Fukuyama congenital muscular dystrophy) [10–15]. These mechanisms potentially contribute to baseline smooth and striated muscle dysfunction in the gastrointestinal tract which may progress to an intestinal pseudoobstructive baseline state, a known precondition for volvulus formation [8,9,15]. While fiber, stool softeners, prokinetics (cisapride/erythromycin), and laxatives, remain the mainstay of maintenance treatment for this aspect of myotonic dystrophy, hope