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Case and Review

Sigmoid Volvulus in Myotonic Dystrophy Type I (Steinert Disease)

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Keywords
Steinert disease · Myotonic dystrophy · Sigmoid volvulus

Abstract
Myotonic dystrophy (MD) is a progressive multisystem genetic disorder that is characterized by progressive muscle weakness and wasting. MD1 (also known as Steinert disease) is associated with various clinical entities such as skeletal muscle weakness, myotonia, cardiac abnormalities, respiratory dysfunction, gastrointestinal involvement, and cognitive impairment. In this case report, we present a 32-year-old woman with MD1 who presented with a sigmoid volvulus, which was treated with endoscopic decompression.

Introduction

Myotonic dystrophy (MD) is a progressive multisystem genetic disorder that is characterized by progressive muscle weakness and wasting [1]. There are two major types of MD that can be distinguished based on clinical and molecular characteristics: myotonic dystrophy type I (MD1), also known as Steinert disease, and myotonic dystrophy type II (MD2) or proximal myotonic myopathy [1]. The prevalence of MD is approximately 1 in 7.400–10.700 and is most common in adults of European ancestry [2–4]. MD1 is caused by the expansion of an unstable cytosine-thymine-guanine (CTG) trinucleotide repeat sequence in the 3′-untranslated region of the myotonic dystrophy protein kinase gene located on chromosome 19q13.3 [5]. Wild-type individuals have between 5 and 34 CTG repeats at this locus, whereas alleles that

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are associated with clinical signs occur with greater than 50 CTG repeats. The CTG repeat expansion usually increases from one generation to another and is moderately correlated with disease severity and age of onset [6–8]. MD1 is associated with various clinical entities such as skeletal muscle weakness, myotonia, cardiac abnormalities, respiratory dysfunction, gastrointestinal involvement, and cognitive impairment [1, 9, 10]. In this case report, we present a woman with MD1 who presented with a sigmoid volvulus.

Case

Here, we present a 32-year old Dutch woman with no medical history, who was admitted to our tertiary center for endoscopic decompression of a sigmoid volvulus. Her facial appearance showed (Fig. 1) characteristic myopathic features with bilateral ptosis, open mouth, and wasting of temporalis and masseter muscles. Neurological exam revealed symmetrical weakness of facial musculature with reduced ability to raise eyebrows and to puff out both cheeks. Furthermore, opening and closing of the fists induced myotonia of the finger extensors, and percussion induced myotonia of the thenar eminence (video). In her family history, the patient had a father and a brother who were diagnosed with MD1; however, she was never seen by any physician. Her father died of cardiopulmonary complications of MD1. The course of admission is illustrated in Figure 2. After endoscopy, the patient became increasingly dyspneic, and her electrocardiogram showed abnormalities (Fig. 3). D-dimer (3,599 ng/mL, normal <500 ng/mL) and NT-Pro BNP (1,242 ng/L, normal <247 ng/mL) were raised. However, CT angiography and cardiac ultrasound did not show significant abnormalities. The patient suffered from abdominal pain, the CRP markedly increased (166.9 to 464.7 mg/L), and CT

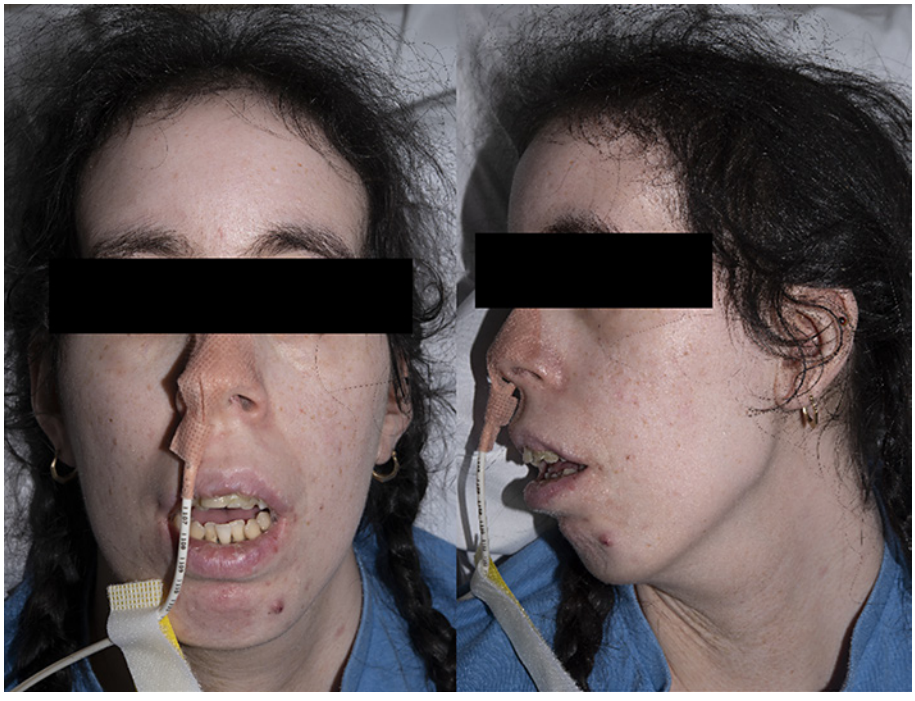

Fig. 1. Facial features of characteristic for myotonic dystrophy of the case reported here. The face is long and the palate is high arched. The cheeks are hollowed and the jaw sags. The patient gave a permission for the publication of the photograph.

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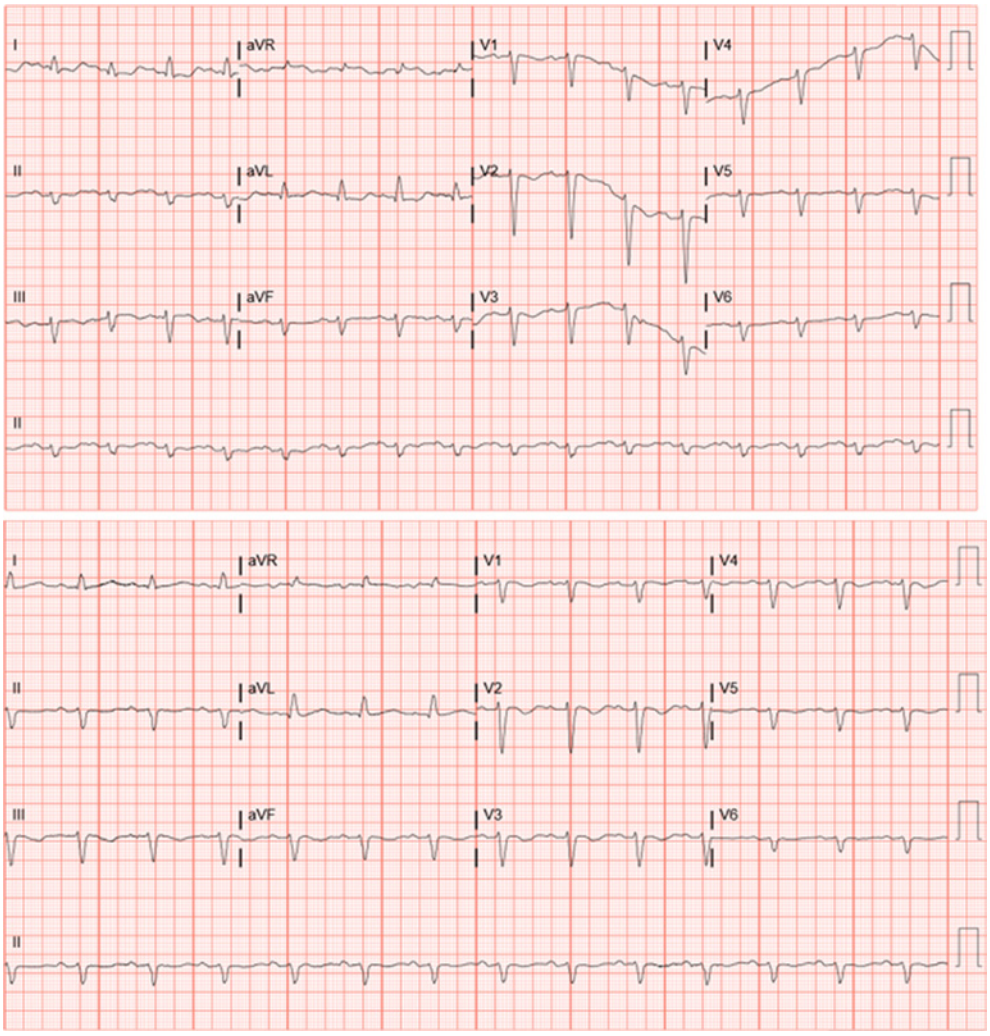

Fig. 2. Course of CRP level during the entire duration of admission. On top of the figure, various events (e.g., CT, drain placements, etc.) were placed in chronical order.

abdomen showed diffuse fluid collections in the pelvis (see Fig. 4). Antibiotic treatment was started with cefuroxime 750 mg 3 times per day and metronidazole 500 mg 3 times per day. A drain was placed in these fluid collections, which were flushed with NaCl 0.9% 3 times a day. However, due to low drain production, the drain was repositioned under ultrasound a few days later. After repositioning, the fluid collections were drained slightly, but were still present; therefore, an additional drain was placed. The drains were then flushed by using alteplase 5 mg in 15 mL NaCl 0.9%, which were rinsed with 10 mL NaCl 0.9%. This flushing strategy was used for 3 days. This has significantly improved drain production, which was shown on follow-up CT that revealed that the collections were fully drained. The CRP markedly decreased (see Fig. 2), clinical symptoms improved (slight abdominal pain, no fever), and antibiotic treatment was ceased. Hereafter, the patient was discharged in a stable clinical condition. During the entire admission, the nutritional intake of this patient was poor due to nausea and abdominal pain. The patient received nutritional support through a feeding tube, and a dietician was involved during admission. She suffered from low levels of iron, potassium, phosphate, and vitamin D, most likely caused by poor intake combined with chronic diarrhea. The patient received suppletion therapy of iron, potassium, and phosphate during admission, and after

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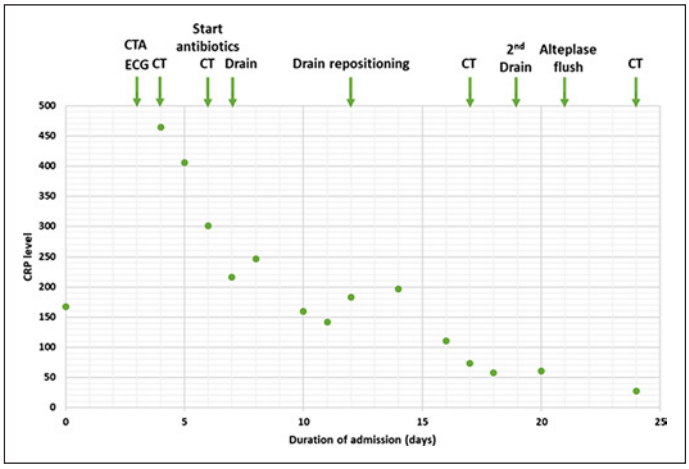

Fig. 3. ECG abnormalities that were found during admission. The ECG shows sinus rhythm, left-sided axis, PR 201 ms, QRS 125 ms on left bundle branch block and anticus block, moderate r-peak progression on anterior wall, and negative diffuse t-peak. ECG, electrocardiogram.

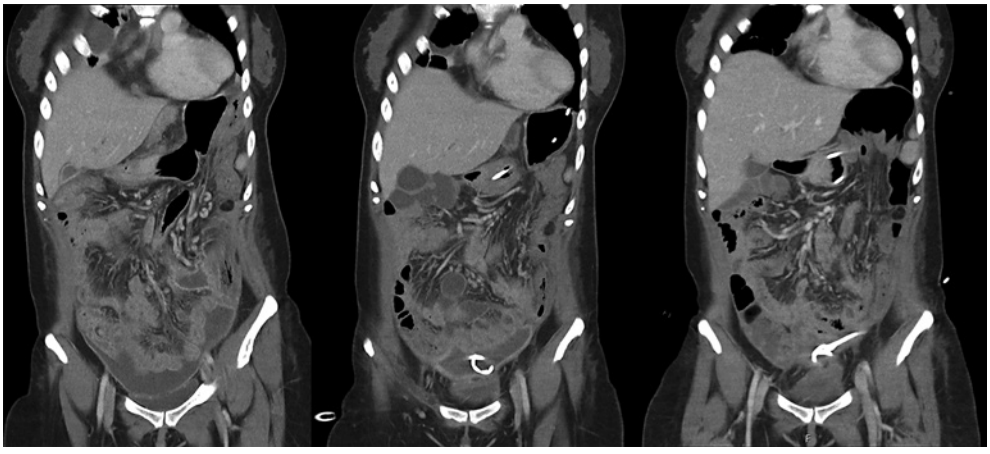

Fig. 4. CT scans of the fluid collections in the pelvis, which are taken on various moments during admission. The CT scans were made on days 6, 17, and 24 of admission. The fluid collections were fully drained on day 24.

discharge, the patient began vitamin D suppletion therapy. Furthermore, the patient also suffered from candidiasis of the tongue, which was successfully treated with nystatin. After discharge, the patient was referred to multiple specialists (i.e., neurologist, cardiologist, gastrointestinal surgeon, ophthalmologist, pulmonologist, clinical geneticist, and oral- and maxillary surgeon) for follow-up of MD1. In addition, the patient will receive support from a dietician and home help provision. The gastrointestinal surgeon will consider a sigmoid resection; this will be discussed in shared decision making with the patient during an outpatient department visit.

Discussion

This case report presents an MD1 patient who underwent endoscopic decompression for a sigmoid volvulus. The patient suffered from fluid collections in the small pelvis, which may have been due to microperforations caused by focal ischemia resulting from the volvulus.

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Table 1. Summary of literature concerning MD and volvulus [24–27]

Author	Age	Sex	MD type	Location	Decompression	Surgery
This case	32	Female	MD1	Sigmoid	Yes	To be determined
da Silva et al. [16]	16	Female	MD1, congenital	Transverse colon	Yes	Extended right hemicolectomy
Mercado-Deare et al. [24]	7	Male	MD?	Transverse colon	Yes	NA
Simpson Khilnani [17]	22	Male	MD?	Sigmoid	Yes	Sigmoid resection
Simpson Khilnani [17]	27	Male	MD?	Sigmoid	No	Sigmoid resection
Brunner et al. [25]	32	Female	MD?	Sigmoid	No	Laparotomy Sigmoid resection
Kusunoki et al. [18]	57	Female	MD?	Stomach	No	Gastrectomy
Kark Greenstein [26]	NA	NA	MD?	Sigmoid	NA	NA
Bertrand [27]	47	Male	MD?	Sigmoid	NA	NA

NA, not available; MD?, myotonic dystrophy without known subtype.

The fluid collections were successfully drained using alteplase flushing, which is more commonly used for intrapleural collections [11]. Alteplase flushing successfully aided in drainage of the pelvic fluid collection and therefore prevented the need for surgical intervention. During admission, it was found that the patient had various deficiencies (e.g., iron, vitamin D), which were supplemented. Nutritional deficiencies have been described in the literature in MD patients [12]. It is therefore important that patients should be followed up by a dietician for assessment of nutritional status. Furthermore, electrocardiogram showed abnormalities (Fig. 3) that prompts follow-up. Cardiac arrhythmias have been commonly described in MD, and the risk for sudden cardiac death is also increased in MD patients [13, 14].

Literature

In the literature, various cases have been described of megacolon, ileus, and volvulus in patients with MD. The summary of literature that has been published on the occurrence of volvulus in MD can be found in Table 1. Yoshida et al. [15] performed a histological study on MD patients in whom a hemicolectomy was performed for megacolon. They found normal smooth muscle histology, but did also report abnormalities (e.g., reduced number of neurons and degenerative neuronal changes) of the myenteric plexus. This indicates a possible neuropathic origin of intestinal dysmotility. Furthermore, de Silva et al. [16] described a 16-year-old female with congenital MD who underwent operative repair for transverse colonic volvulus. This case presented with severe, sharp, left lower quadrant, epigastric, and periumbilical, nonradiating abdominal pain for 5 days. She was also known to have chronic obstipation. Simpson and Khilnani [17] reported 2 patients with MD that had sigmoid volvulus. The first patient was a 22-year-old male, who was diagnosed with MD during childhood, who suffered from severe recurrent crampy abdominal pains associated with either diarrhea or black stools occurring 1–3 times per week and occasionally lasting 1–2 days. This patient had a sigmoid volvulus that was reduced with sigmoid intubation. The second patient was a 27-year-old male with MD, who had recurrent episodes of colicky periumbilical pain associated with black stools and diarrhea. Barium enema revealed a sigmoid volvulus, which was treated an elective sigmoid resection. Furthermore, Kusunoki et al. [18] reported a gastric volvulus in a 57-year-old female with MD who underwent

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emergency gastrectomy. This patient presented with sudden abdominal pain, nausea, and vomiting. Also, Sasaki et al. [19] reviewed the literature on the association of sigmoid volvulus with megacolon in MD. They described that in all patients with a megacolon in the sigmoid, it was preceded by a sigmoid volvulus. Furthermore, recurrence rates of a sigmoid volvulus are high (approximately 54–86%) [20–22], and it is not known how high the recurrence rate is in MD1 patients in particular. It can be hypothesized that patients with MD1 are more suspectable for higher recurrence rates due to intestinal dysmotility that might play a role in these patients [15]. Furthermore, a sigmoid volvulus could lead to ischemia, perforation, and mortality [23]. Hence, this would be an argument to plan an elective sigmoid resection in MD patients who suffered from a sigmoid volvulus.

Conclusion

MD1 is a rare multisystem progressive genetic disease, which is associated with gastrointestinal symptoms. A sigmoid volvulus can occur with MD1 (Steinert disease), which requires adequate (surgical) treatment. In case of postprocedural pelvic fluid collections, alteplase flushing may be useful. Physicians should be alert in patients suspect for MD. It is very important that these patients are followed up by multiple clinical specialists.

Statement of Ethics

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images. According to Dutch National Guidelines, there is no requirement for Ethics Approval by the Medical Ethical Review Committee in the Netherlands (https://www.rijksoverheid.nl/documenten/rapporten/2020/02/14/niet-wmo-plichtig-onderzoek-en-ethische-toetsing, assessed August 15, 2021).

Conflict of Interest Statement

The authors of this manuscript do not have any conflict of interest to declare.

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Author Contributions

A.B.B.: manuscript writing, creation of figures, and editing; J.S.G.: neurological exam and critically reviewing manuscript; and M.E.T.: critically reviewing manuscript.

Data Availability Statement

All data generated or analyzed in this case report are included in the article.

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