Respiratory Failure Secondary to Pseudo-obstruction in a Patient with Myotonic Dystrophy Type 1

Jordan Earl PA-S, Sheree Piperidis, PA-C
Quinnipiac University Graduate Physician Assistant Program

Introduction

- Myotonic dystrophy (DM) is an inherited, autosomal dominant myopathy resulting in progressive musculoskeletal weakness. It is caused by expansion of cytosine-thymine-guanine (CTG) repeat in the 3'-untranslated region of the DM1 protein kinase (DMPK) gene on chromosome 19q13.3.[1]
- Affecting 0.5 -18.1 per 100,000 individuals, it is one of the most common causes of muscular dystrophy in adults.[2]
- Myotonic dystrophy can have complications which affect several body systems.[1]
- Respiratory failure is one of the most common causes of death in patients with type 1 myotonic dystrophy (DM1), likely due to both respiratory drive factors and skeletal muscle weakness.[3]
- This disorder has also commonly been associated with gastrointestinal manifestations, such as gastroesophageal reflux, dysphagia, cholelithiasis, and intestinal dysmotility.[1]
- Pseudo-obstruction has also been described as a rare complication of DM in few published cases.[4,5]
- Chronic intestinal pseudo-obstruction is an uncommon disorder which is characterized by abnormalities affecting smooth muscular contractions of the gastrointestinal tract and can lead to signs and symptoms consistent with an intestinal obstruction in the absence of a mechanical cause.[6]
- Pseudo-obstruction/gastric dilatation can lead to acute dyspnea in rare cases and may be improved dramatically with bowel decompression.[7]

Table 1. Complications of Myotonic Dystrophy by Body System[1]

Cardiovascular	• Atrial fibrillation • Conduction defects • Dilated cardiomyopathy
Pulmonary	• Neuromuscular-related respiratory failure
Central Nervous System	• Intellectual disabilities • Cerebrovascular accident • Anxiety and depression • Hypersomnia and sleep apnea
Ophthalmologic	• Cataracts
Gastrointestinal	• Dysphagia • Gallstones, cholecystitis • Transaminitis, liver enzyme elevations • Intestinal pseudo-obstruction
Endocrine	• Insulin insensitivity • Male infertility
Musculoskeletal	• Progression loss of motor function • Myalgias • Impairments in ADLs due to distal muscle weakness
Dermatologic	• Androgenic alopecia • Basal cell carcinoma • Pilomatrixomas
Gynecological	• Increased risk of spontaneous abortion, placenta previa, pre-term birth, prolonged labor, dysmenorrhea

Case Description

Brief History

- 40-year-old Hispanic female
- Past medical history:
  - Myotonic dystrophy type 1 (DM1)
  - GERD
  - IBS
  - Asthma
- Patient presented to the emergency department with a chief complaint of generalized weakness for the past 5 days.
- Also noted having a "tingling" sensation in her upper extremities bilaterally, "heavy" sensation in her lower extremities bilaterally, and mild difficulty breathing . Endorsed one episode of non-bloody diarrhea 4 days prior to presentation.
- Denied cough, fever, chills, headache, abdominal pain. No sick contacts.
- Surgical history: cesarean section, cholecystectomy
- Medications: norethindrone 0.35 mg po daily, albuterol 1-2 puffs q4 hours prn
- Allergies: none
- Family and social history unremarkable.
- Figure 1 describes patient's hospital course.

Objective Findings

- Vitals: BP: 127/70 mm/Hg, afebrile, PO_2 78% on BiPAP
- General appearance: Acutely ill, lethargic
- Lungs: Clear lung sounds bilaterally, exam limited due to patient on BiPAP.
- Heart: Normal rate and regular rhythm.
- Abdomen: Distended. Soft, non-tender. Normal active bowel sounds. No hernias or palpable masses. No hepatosplenomegaly. No rectal masses or bleeding.
- MSK: Bilateral lower extremity edema.
- Neurological: Unable to follow commands, minimally responsive to noxious stimuli.

Diagnostic Findings

- ABG: pH 7.21, CO2 >100, O2 72
- Urine tox was negative.
- Respiratory viral panel was negative.
- No leukocytosis, no anemia.
- AST: 225, ALT: 550
- Imaging below (Figures 2 and 3)

Fig. 1 Hospital Course

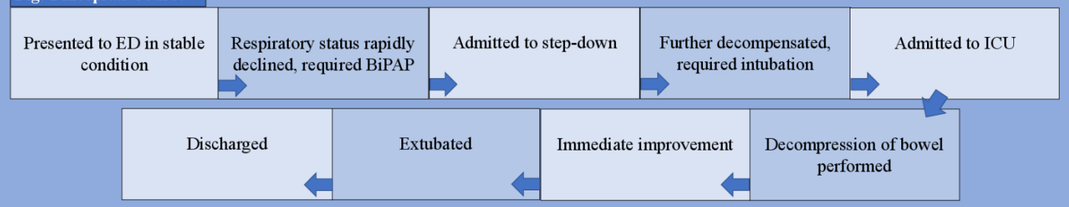
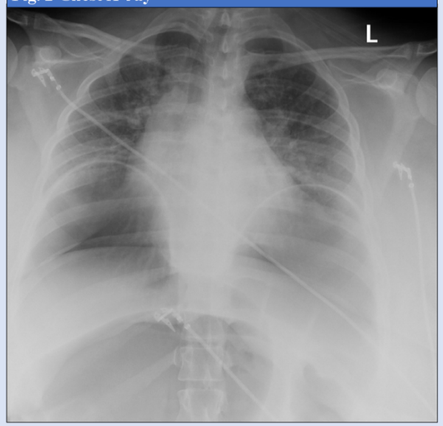


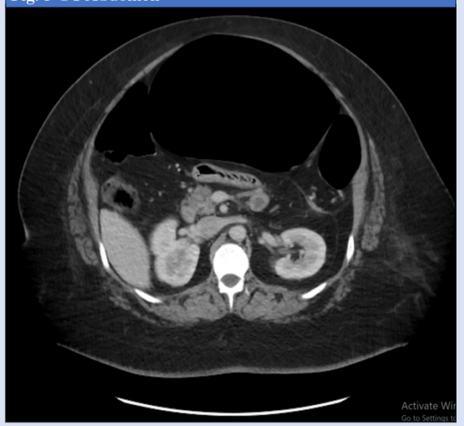
Fig. 2 Chest X-ray



Diagnostic Imaging

- XR chest was significant for hypoexpanded lungs, hazy left hilar opacity, prominent appearance of subdiaphragmatic lucencies, corresponding with severely dilated colon (Figure 2).
- CT abdomen pelvis was significant for markedly gaseous distention of the transverse colon, without obstruction, measuring 17 mm in its greatest dimension (Figure 3).

Fig. 3 CT Abdomen



Management and Outcome

- The patient was intubated and transferred to the ICU for further management and monitoring.
- Gastroenterology was consulted for colonic pseudo-obstruction and performed bowel decompression resulting in significant improvement in abdominal tension and girth.
- The patient's condition improved immediately after the procedure, and she was successfully extubated the following day. Mentation returned to baseline.
- Neurology was consulted to evaluate the possibility of myotonic dystrophy as a cause for her respiratory failure.
- Patient was transferred to floor level of care on 2 L O2 and then weaned to room air.
- Patient was monitored for several days and discharged home in stable condition on day 6.
- Able to tolerate normal diet and normal bowel movements.
- Patient was assessed by PT and recommended home services.
- Outpatient follow-up with neuromuscular clinic to further manage DMI.
- Outpatient follow-up with gastroenterology to monitor for recurrence of pseudo-obstruction/colonic dilatation.

Conclusion

- Respiratory failure is one of the most common causes of death in patients with DM1.
- Although a rare complication of myotonic dystrophy, pseudo-obstruction/colonic dilatation should be considered in the differential diagnosis of a patient with DM presenting with respiratory distress.
- Prompt decompression can lead to an immediate improvement in the patient's condition.

References

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