Pneumonia and de novo atrial fibrillation in a patient with myotonic dystrophy type 1
A case report
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Abstract
Introduction: Myotonic dystrophy type 1 is a rare genetic disorder that mainly affects the musculoskeletal system; However, it may cause several complications in other body systems representing challenges for health care providers.

Patient concerns: We present the case of a patient with a history of previously diagnosed type 1 myotonic dystrophy who presented to the emergency department with cough dyspnea, and thoracic pain.

Diagnosis: Differential diagnoses included pulmonary embolism with a moderate probability according to the Wells score, acute coronary syndrome, acute heart failure, and pneumonia. Diagnostic workup involved chest radiography, EKG, and a CTPA which revealed pneumonia, posteriorly the patient presented de novo atrial fibrillation.

Interventions: The patient was successfully treated with empiric antibiotic therapy and amiodarone, respiratory and physical therapy.

Outcomes: The patient was discharged on day 34, however oxygen weaning was not possible.

Conclusion: Treatment of MD1 patients is challenging due to the various mechanisms of the disease; patients with new-onset deterioration should be screened for the most common complications such as cardio-respiratory events. The authors suggest pneumonia as a risk factor for basal respiratory function deterioration and a contributing factor for triggering cardiac events for further research in prospective studies.

Abbreviations: AF = atrial fibrillation, BMI = body mass index, CRP = C-reactive protein, CTG = cytosine, guanine, thymine, CTPA = computed tomography pulmonary angiogram, EKG = electrocardiogram, ER = emergency room, FVC = Forced Vital Capacity, IAB = interatrial block, ICU = intensive care unit, MD1 = myotonic dystrophy type 1, NIV = non-invasive ventilation, NRB = non-rebreather mask.

Keywords: atrial fibrillation, case report, Myotonic dystrophy type 1, pneumonia

1. Introduction
Myotonic dystrophy type 1, also known as Steinert disease, is the most common type of dystrophy. It is an autosomal dominant triplet-repeat expansion disorder that affects between 1:3000 and 1:8000 individuals worldwide. Pulmonary complications are the leading cause of death in such patients. Cardiac complications are the second leading cause of death in myotonic dystrophy type 1; The most common cardiac pathologies are arrhythmias, including heart block, atrial fibrillation and flutter, sinus bradycardia, and ventricular tachycardia.[1,2]

Currently, there are few management guidelines on inpatient care for these patients, such as the consensus-based care recommendations for adults with myotonic dystrophy type 1.

We report a diagnostic, therapeutic approach, and complications of a patient with myotonic dystrophy presenting to the ER with cough, dyspnea, and thoracic pain.

2. Case presentation
A 36-year-old woman was admitted to the emergency department with a history of cough per 4 days with associated whitish expectoration, dyspnea, and thoracic pain.

Her medical history included myotonic dystrophy type 1, obstructive sleep apnea, hypothyroidism, ischemic cardiomyopathy, and obesity. A traumatic history revealed a left Weber B fracture 3 months before admission. Surgical history included appendicectomy and cholecystectomy. Her family medical history myotonic dystrophy type 1 and ischemic cardiomyopathy in her older brother.

On examination, she was hypoxic in room air, and 92% oxygen saturation was achieved with a non-rebreather oxygen mask at 50% FiO2. In addition, she was tachycardic (105/bpm), poly-pneic (28/rpm), and afebrile. Anthropometric data on admission revealed a BMI of 30.4 kg/m2.
Cardiopulmonary examination revealed a regular cardiac rhythm, bilateral crackles at the lung bases, and 2+ pitting edema in her lower extremities. Arterial blood gases showed exacerbated chronic respiratory acidosis with hyperlactatemia (1.2 mmol/L) and PaO/FiO ratio of 132.34. D dimer was elevated at 6.5 mg/dL. The basic metabolic panel and liver function test results were within normal limits. T-troponin levels were also elevated at 0.027. A 12-lead EKG showed a left-deviated axis with an anteroseptal injury. Chest radiography (Fig. 1) demonstrated moderate global cardiomegaly and interstitial opacities disseminated in the bilateral lung parenchyma, with concerns for atelectasis vs. infiltrative origin. Due to an inconclusive radiography, a CT scan was obtained, which showed multilobar consolidations involving the left upper lobe and both lower lobes (Figs. 2, 3).

Our differential diagnosis included pulmonary embolism with a moderate probability according to the Wells score, acute coronary syndrome, acute heart failure, and pneumonia. The control troponin T level was negative. MRSA cultures and SARS-Cov2 RT-PCR cultures were negative. A CT pulmonary angiography (CTPA) was negative for pulmonary embolism.

After CTPA, the patient clinically deteriorated as she became cyanotic, and had an altered mental status. The patient showed irregular breathing and increased respiratory effort. Her arterial oxygen saturation was 40% on a nasal cannula running at 4 L/min. The oxygen device was promptly changed to a non-rebreather mask (NRB) at a flow rate of 15 L/min. The patient's condition continued to deteriorate, mechanical invasive ventilation was initiated, and she was transferred to the ICU.

Empiric treatment with ampicillin-sulbactam (3 gr every 6 h) and clarithromycin (500 mg every 12 h) was administered for seven days. After five days in the ICU, the film array returned positive results for rhinovirus. Due to a lack of improvement in respiratory parameters, a tracheostomy was performed, and new cultures were obtained, which were negative. The patient continued to receive respiratory rehabilitation and was subsequently transferred to a medical ward. She was extubated and continued with a tracheostomy mask but presented signs of increased respiratory effort, low oxygen saturation, and tachycardia (200 bpm). An EKG was obtained, which showed atrial fibrillation with a rapid ventricular response. Based on these findings, two boluses of amiodarone (150 mg) were administered.

Due to the acute clinical changes, respiratory rehabilitation was continued, but with very tight parameters for oxygen weaning. Pulmonology was consulted, and they indicated that short-term removal of the tracheostomy might not be possible because respiratory compromise was due to the underlying disease. The patient was discharged on day 34 with tracheostomy with 30% FiO2 and maintaining oxygen saturation at 91%.

3. Discussion
Myotonic dystrophy type 1 is the most common muscular dystrophy in adults and is associated with early mortality with an average age of 53 years. The main cause of death is respiratory failure (43% of cases), followed by cardiovascular disease (20%) and sudden death (11%), presumably due to cardiac arrhythmias.[2]

Respiratory compromise in these patients has been proposed to be a result of myotonia of the inspiratory and expiratory muscles, which results in a less compliant chest wall and manifests as increased breathing work, decreased tidal volume, and CO2 retention.[3] Recently, the factors influencing the progression of respiratory dysfunction have been studied. Hartog et al. conducted a retrospective study of 110 adult patients seen in the Neurology Department at Ohio State University and found numerous factors influencing respiratory declination parameters measured by FVC, including greater CTG repeat sizes, muscular impairment rate scale (MIRS), BMI, and the presence of respiratory symptoms such as orthopnea, shortness
of breath, dyspnea on exertion, and NIV compliance. Overall, respiratory muscle impairment was associated mostly with respiratory symptoms. Additionally, they found a mean rate of −1.42% decline in FVC per year, independent of age.\(^4\) There are no studies in the current literature that establish a relationship between pneumonia in MD1 patients as a risk factor for deterioration of respiratory function or the requirement for an increase in oxygen supply in NIV. The incidence of cardiac complications in these patients develops because of many interacting factors. A study conducted by Antonini et al reported the natural history of cardiac involvement in patients with myotonic dystrophy type 1. This prospective study included 50 patients who underwent periodic EKG and Holter monitoring during a median follow-up of 56 months. It showed that 38% of this cohort developed major EKG changes; of these, 9 subjects developed major conduction changes, including atrioventricular blocks and bundle-branch blocks, 5 developed major arrhythmias, consisting of ventricular premature complexes (1), supraventricular tachycardia (ST), ventricular premature complexes (1), and atrial fibrillation (AF) (3), of which only 1 was symptomatic. The remaining 5 patients developed major cardiac abnormalities in conjunction with cardiac arrhythmias.\(^3\) Other studies have suggested factors influencing the development of atrial fibrillation; Russo et al. enrolled 70 MD1 patients who underwent pacemaker implantation for cardiac rhythm abnormalities for a minimum of 2-year follow-up, analyzed EKGs, and found interatrial block (IAB) in 22 patients. During the follow-up, AF episodes were detected in 18 patients.\(^6\)

Second, in non-MD1 patients, the incidence of cardiovascular complications after pneumonia has been discussed; a systematic review found a 4.7% incidence of arrhythmias in the first month after community-acquired pneumonia.\(^7\) Acute inflammatory processes, such as respiratory infections, can increase large artery stiffness and pulse wave reflections from the middle and small arteries, increasing the left ventricular afterload. Furthermore, they can increase pulmonary arterial pressure and, therefore, raise the right ventricular afterload.\(^8\) All these processes are of great importance in patients with established cardiac diseases, such as MD1, who may deteriorate their basal status in the context of infectious diseases.

Management of pneumonia and cardiovascular complications in patients with MD type 1 is challenging, largely owing to the complex mechanisms of the disease.

This case report describes the diagnostic and therapeutic approach given to a patient with MD1 presenting to the ER. This highlights the importance of annual screening with EKG because of the high incidence of cardiovascular complications and early mortality. Additionally, we propose pneumonia as a risk factor for basal respiratory function deterioration and a contributing factor for triggering cardiac events for further research in prospective studies.

**Author contributions**

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