Case Report

Mediastinal and pleural lipomatosis as a manifestation of myotonic dystrophy type 1

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ABSTRACT

Mediastinal and pleural lipomatosis is a rare but usually benign and asymptomatic disease. Mediastinal lipomatosis is associated with steroid use, obesity, hyperlipidemia, diabetes, or Cushing syndrome. In some cases, it becomes symptomatic manifesting with dyspnea, thoracic pain, coughing, dysphonia, dysphagia, supraventricular tachycardia, or persistent pneumonia. Mediastinal lipomatosis has not been reported in association with myotonic dystrophy type 1 (MD1). In a 65yo male with a long-term history of progressive muscle weakness, hyper-creatine-kinase-emia, bilateral cataract, sleep apnea syndrome, gynecomastia, hepatic steatosis, arterial hypertension, atrioventricular block 1, QTc prolongation, hyperlipidemia, hyperuricemia, and hepatopathy, MD1 was diagnosed upon the clinical presentation and a heterozygous CTG repeat expansion of 1200–1400 repeats in *DMPK*. Work-up for dyspnea and leg edema revealed heart failure and mediastinal and pleural lipomatosis. Upon standard treatment, heart failure resolved. In conclusion, mediastinal and pleural lipomatosis can be a rare manifestation of MD1 and can manifest with heart failure. In patients with mediastinal lipomatosis, MD1 should be excluded.

KEY WORDS: Cardiac involvement, CTG repeats, lipomatosis, multisystem, myotonic dystrophy

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Submitted: 13-Jan-2021 **Accepted:** 10-Feb-2021 **Published:** 30-Aug-2021

INTRODUCTION

Myotonic dystrophy type 1 (MD1) is the most prevalent myopathy in adults and due to a CTG repeat expansion >50 repeats in *DMPK*.[1] The phenotypic spectrum of MD1 is highly variable and depends on the mutation load (homozygote and heterozygote), the CTG repeat expansion size, and the disease stage,[1] The most common phenotypic features include myopathy, myotonia, cataract, frontal baldness, atrioventricular (AV) block 1, cardiomyopathy, hypogonadism, cognitive impairment, and pilomatricoma.[1] Disease severity is categorized as mild, classic, or congenital.[1] Mediastinal and pleural lipomatosis has not been reported as a phenotypic feature of MD1.

CASE REPORT

The patient is a 58-year-old Caucasian male, height 178 cm, weight 100 kg, with a previous history of smoking until age 49 years, tuberculosis, slowly progressive muscle weakness, bilateral cataract, sleep apnea syndrome, gynecomastia, recurrent nonspecific, thoracic pain, arterial hypertension, hepatic steatosis, AV block 1, QTc prolongation, hyperlipidemia, hyper-creatine-kinase-emia,

Access this article online

Quick Response Code:

Website: www.lungindia.com

DOI: 10.4103/lungindia.lungindia_25_21

How to cite this article: Finsterer J, Scorza FA. Mediastinal and pleural lipomatosis as a manifestation of myotonic dystrophy type 1. Lung India 2021;38:486-8.

hyperuricemia, elevated myoglobin, and hepatopathy. At age 36 years, classical MD1 was diagnosed upon the typical clinical presentation and myogenic needle electromyography. At age 51 years, echocardiography revealed markedly increased pericardial fat. At age 52 years, a computed tomography (CT) scan of the thorax revealed mediastinal lipomatosis. At age 53 years, he was admitted because of dyspnea and leg edema. Genetic work-up revealed a heterozygous CTG repeat expansion of 1200–1400 repeats in *DMPK*. On X-ray of the lungs, marked pleuropericardial adhesions were detected and a CT scan of the thorax revealed an esophageal diverticulum and confirmed marked mediastinal and moderate pleural lipomatosis [Figure 1]. Standard therapy resolved heart failure. The patient was discharged with acetylsalicylic acid, allopurinol, doxazosin, famotidine, furosemide, and simvastatin. His further course was progressive as he developed diabetes, dysphagia, and ultimately died at age 58 years.

DISCUSSION

The patient is interesting for the presence of mediastinal and pleural lipomatosis, which has not been reported in MD1. Mediastinal lipomatosis is usually a benign and asymptomatic condition, characterized by the accumulation of fatty tissue within the mediastinum or pleura.[2] In single cases, it may clinically manifest with dyspnea,[3] thoracic pain, cough, dysphonia, dysphagia, supraventricular tachycardia,[4] or persistent pneumonia.[5] Mediastinal lipomatosis associated with pleural lipomatosis is more likely symptomatic than mediastinal lipomatosis alone. Pleural lipomatosis can be mixed up with pleural effusion.[6] and mediastinal lipomatosis with cardiomegaly.[7] In rare cases, mediastinal lipomatosis causes tracheal stenosis[8] or compression of the right ventricular outflow tract.[4] Mediastinal lipomatosis may lead to low-voltage electrocardiogram[9] or hemomediastinum.[10] The cause of mediastinal lipomatosis is unclear, but it has been previously reported in association with the use of steroids,[11] obesity,[2] diabetes,[3] Cushing syndrome,[3] hyperlipidemia,[3] surgery,[12] or sarcoidosis.[11]

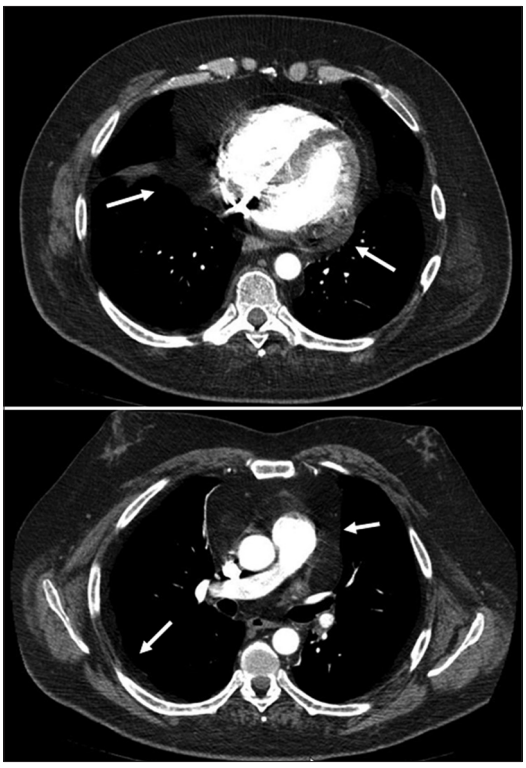

Figure 1: Computed tomography scan of the thorax showing marked mediastinal and moderate pleural lipomatosis (arrows)

The presented patient did not take steroids and initially had no diabetes. He was never tested for Cushing syndrome, but his clinical presentation did not suggest it. Clinically, there was no indication for ectopic adrenocorticotropic hormone or steroid production. Thus, the only risk factors for mediastinal lipomatosis present in the index patient were hyperlipidemia and obesity. However, since millions of people have dyslipidemia or obesity without developing mediastinal lipomatosis, mediastinal lipomatosis is more likely due to MD1 than due to hyperlipidemia. Whether mediastinal lipomatosis or cardiomyopathy was responsible for heart failure remains speculative. Heart failure in association with mediastinal lipomatosis has been reported only once,[13] but this patient also had dilated cardiomyopathy and lupus erythematosus.[13] An argument against heart failure as the cause of dyspnea and leg edema is that proBNP was repeatedly only slightly elevated.

In conclusion, the presented case shows that mediastinal and pleural lipomatosis can be a rare manifestation of MD1, that it may manifest with heart failure, and that standard heart failure treatment resolves symptoms and signs completely. In patients with mediastinal lipomatosis, MD1 should be excluded.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given his consent for his images and other clinical information to be reported in the journal. The patient understands that his name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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