Case Report

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

Josef Finsterer¹, Fulvio A Scorza²
¹Municipal Hospital Landstrasse, Messerli Institute, Vienna, Austria, ²Department of Neuroscience, University of Sao Paulo, São Paulo, Brasil

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

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 pilomatrixcoma.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, hepatic steatosis, arterial hypertension, atrioventricular block 1, QTc prolongation, hyperlipidemia, hyper-creatine-kinase-emia.
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 \textit{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,\(^2\) hyperlipidemia,\(^3\) surgery,\(^12\) or sarcoidosis.\(^11\)

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.

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**Conflicts of interest**

There are no conflicts of interest.

**REFERENCES**

1. Bird TD. Myotonic Dystrophy Type 1. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJ, Stephens K, et al. editors. GeneReviews\(^\text{®}\). Seattle (WA): University of Washington; 1999-2021.
2. Lim JY, McAnulty KA, Chang CL. Dyspnoea and restrictive lung disease due to mediastinal and pleural lipomatosis in morbid obesity. Respirol Case Rep 2019;7:e00421.

3. Singh A. Mediastinal lipomatosis with dyslipidemia: Cause of dyspnea. J Assoc Physicians India 2016;64:88-9.

4. Bulakci M, Yahyayev A, Ucar A, Erer B, Erer B, Dunsun M. Unusual cause of right ventricular outflow tract compression: Mediastinal lipomatosis. J Thorac Imaging 2011;26:W134-6.

5. Concepcion E, Amadio J, Lee H. Mediastinal lipomatosis presenting as persistent pneumonia. J Pediatr 2015;167:493-e1.

6. Kaur N, Singh J, Haq S, Garg S, Bhatnagar S. Pleural and mediastinal lipomatosis with subpleural fat as a mimic of pleural effusion: A rare case report. J Clin Diagn Res 2017;11: TD03-4.

7. Laakso M, Suhonen M, Helin M. Mediastinumin lipomatoosin aiheuttama sydänvarjon suureneminen [Mediastinal lipomatosis simulating cardiomegaly]. Duodecim 1984;100:96-9.

8. D’Andrilli A, Vanni C, Venuta F, Rendina EA. Critical tracheal stenosis caused by mediastinal lipomatosis: Long-term efficacy of airway stenting. J Thorac Cardiovasc Surg 2015;149:e109-10.

9. Puttarajappa C, Dhoble A. Mediastinal lipomatosis as a cause of low voltage complexes on electrocardiogram and widened mediastinum: A case report. Cases J 2008;1:171.

10. Taillé C, Fartoûkh M, Houël R, Kobeiter H, Rémy P, Lemaire F. Spontaneous hemomediastinum complicating steroid-induced mediastinal lipomatosis. Chest 2001;120:311-3.

11. Qiu JK, Dwivedi A, Alter E, Halpern D, Katz ES, Donnino R, et al. A rare case of sarcoidosis-induced polyserositis and steroid-induced mediastinal lipomatosis masquerading as an epicardial tumor. CASE (Phila) 2020;4:166-9.

12. Gudjoncik A, de Carvalho A, Loris L, Chavent A, Cercueil JP, Jazayeri S, et al. Postoperative mediastinal lipomatosis. Arch Cardiovasc Dis 2008;101:593-4.

13. Salerno G, Carbone A, Rea G, Valente T, D’Andrea A, Di Maio M, et al. Unexpected transesophageal echocardiography tee finding: Mediastinal lipomatosis fakes an aortic intramural haematoma. Quant Imaging Med Surg 2017;7:149-51.