Annals of Medical and Clinical Case Reports
Volume 1| Case Report

A Case of Myotonic Dystrophy Type 1 Initially Presenting As Dilated Cardiomyopathy

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Citation: Shannon Cowie, A Case Of Myotonic Dystrophy Type 1 Initially Presenting As Dilated Cardiomyopathy., Ann Med Clin Case Rep, 2026; 1(8): 1-6.

Published Date: 15-11-2025 **Accepted Date:** 13-11-2025 **Received Date:** 07-11-2025

Abstract

Background

Myotonic dystrophy type 1 (DM1) is the most prevalent form of adult-onset muscular dystrophy [1], affecting 10:100,000 across Europe [2]. Cardiac involvement is a major cause of premature death in this population.

Case Summary

A patient presented with breathlessness and was found to have biventricular failure with a negative genetic screen. Three years later they were admitted after a fall due progressive leg weakness and was diagnosed with Myotonic Dystrophy type 1, requiring multi-disciplinary management. This report highlights the patient journey to a unifying diagnosis, focusing on cardiovascular involvement.

Discussion

Despite DM1 being rare, recognising this relationship may enable earlier diagnosis and management.

Learning Objectives

To recognise that, although rare, myotonic cardiac dysfunction can be the first presentation of myotonic dystrophy type 1 (DM1). Myotonic cardiac dysfunction should be considered in those of a younger age with a family history of dilated cardiomyopathy, particularly if their standard genetic panel for dilated cardiomyopathy is normal. The case also highlights that symptoms in patients with DM1 can be multi-factorial and patients with DM1 are therefore best managed by a multi-disciplinary team.

1. Introduction

Myotonic dystrophy type 1 (DM1) is a multi-system genetic disorder affecting skeletal and smooth muscle, with potential involvement of the respiratory, central nervous and cardiovascular systems. It is the most prevalent adult-onset muscular dystrophy, affecting 10:100,000 across Europe [1]. DM1 is caused by an autosomal dominant CTG trinucleotide repeat expansion in the DMPK gene [2]. Cardiac involvement can be seen in around 80% of patients [3] and it is a major cause of premature death. Although rare, DM1 should be considered when assessing cardiac dysfunction to facilitate early diagnosis and management.

2. Case Report

A 58-year-old female was referred to the Cardiology clinic with several months of breathlessness, worse at night, but not specifically on lying flat. The general practitioner (GP) had noted a new left bundle branch block (LBBB) on ECG and an elevated N-terminal pro b-type natriuretic peptide (NTpro-BNP) at 1331 ng/L. She had no past medical history other than anxiety (sertraline and diazepam) but had a family history of cardiomyopathy, with both father and paternal aunt affected. She denied angina or swelling of her legs. She was a non-smoker and only had alcohol in moderation.

On examination, she had a high body mass index (BMI) at 46 kg/m² (weight 126kg, height 166 cm). There was no peripheral oedema, but basal crackles were heard on chest auscultation. Heart sounds and abdominal examination were normal. An ECG in clinic confirmed LBBB with new atrial fibrillation (AF) at 118bpm. Echocardiography showed biventricular dilatation and severe left ventricular systolic dysfunction (ejection fraction <35%). The initial diagnosis was dilated cardiomyopathy secondary to poor AF control.

Standard therapy (Bisoprolol 5mg, Spironolactone 25mg once daily, Ramipril 1.25mg and Furosemide 40mg once daily) for systolic heart failure was trialled although symptomatic hypotension precluded the ongoing use of an ACE inhibitor. Genetic testing for 24 cardiomyopathy-associated genes, including those linked to Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC), was negative. Repeat ECGs showed persistent LBBB and progressive QRS widening (160ms). After rate control with beta-blockers and digoxin (250 mcg), the patient was considered for cardiac resynchronisation therapy (CRT); however, later QRS narrowing (<120ms) made CRT-D unnecessary. Coronary angiography showed non-obstructive plaque disease. Subsequent echocardiograms showed normalisation of LV function.

Several years later, the patient was referred to Neurology with progressive leg weakness and gait instability over a period of three years, now causing frequent falls. Whilst awaiting a routine Neurology appointment, the GP recontacted the team due to progression in the patient's symptoms. The GP had noted worsening balance and a broad-based gait.

Neurological examination revealed bilateral ptosis with eye closure weakness, mild dysarthria, and myotonia on eye opening. Up-gaze was reduced, but without diplopia and visual fields were normal. Tone was also normal. Finger flexion and abduction were Medical Research Council (MRC) Grade 4/5, but proximal upper limb muscles were stronger. Toe and ankle plantar flexion were MRC Grade 3/5 and dorsiflexion was MRC Grade 4/5 with a similar pattern of increased power proximally in the lower limbs. Reflexes were reduced; sensation and coordination were normal.

Neurophysiological testing showed absent peroneal and tibial motor responses, small right ulnar motor responses and normal sensory potentials. Electromyography (EMG) revealed some myopathic looking motor units and clear and infrequent myotonic discharges in upper limb muscles. Creatinine kinase (CK) was elevated at 664U/L. Genetic testing with a polymerase chain reaction (PCR) assay confirmed myotonic dystrophy type 1 with over 100 CTG repeats.

At diagnosis, echocardiography again showed severe LV systolic dysfunction and ECG demonstrated PR prolongation, LBBB and wide QRS. She subsequently received a cardiac resynchronization therapy defibrillator (CRT-D). Spirometry revealed restrictive lung disease (FEV1 57% FVC 54%, FEV1: FVC Ratio 1.05), prompting referral to the Respiratory team.

3. Discussion

This case study demonstrates the non-linear development of multisystem involvements in patients with DM1. The most common cardiac complication arising in patients with DM1 are conduction system defects [4]. Left ventricular dysfunction, as seen in this case, is prevalent only in 7.2-11.3% of patients with DM1 [5]. Other features seen in DM1 are listed in table 1. Although this patient did show signs of conduction system defects on ECGs (PR prolongation and LBBB), they initially presented with symptoms, and preliminary investigations, suggestive of heart failure and were initiated on appropriate treatments for systolic dysfunction. LV dysfunction in this cohort is associated with advanced age, a higher number of CTG repeats, prolonged PR interval and prolonged QRS complex [6].

This case is interesting in that there was no family history of myotonic dystrophy although there was a family history of cardiomyopathy. This condition normally shows genetic anticipation; with more pronounced features seen in those of a younger age and conversely potentially only subtle features in those who present at an older age. It is therefore important to take a full family history and enquire about family history of all the individual features of condition.

Respiratory dysfunction, particularly of a restrictive nature, is common in DM1 and can result in chronic hypercapnia and sleep apnoea [7]. This would be in-keeping with the patient's history as they described breathlessness that was particularly worse at night. It is felt that right ventricular failure in this population generally occurs because of untreated respiratory muscle weakness and

subsequent pulmonary hypertension [8]. This may explain the patient's initial echocardiogram findings which showed severe left ventricular (LV) and right ventricular (RV) impairment.

50% of patients with DM1 are overweight or obese putting them at increased risk of type 2 diabetes [9]. The patient's high BMI was noted but it could be the case that losing weight would have been even more difficult for this patient given the association of this condition with the metabolic syndrome. Interestingly, some studies have now suggested that GLP-1 receptor agonists, previously used in the treatment of type 2 diabetes, may be of benefit in patients with heart failure who are also obese [10].

This case is unusual in that the presenting feature was of cardiac dysfunction, with the classical symptoms of muscle weakness, myotonia and cataracts appearing much later in this case [5]. It is unusual for patients to present with symptomatic heart failure early as normally patients' mobility, and therefore oxygen demand, is limited. Despite DM1 being relatively rare, it is important to recognise the relationship between this condition and cardiac dysfunction to promote earlier diagnosis and treatment.

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