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(CASE REPORT)

Between Strength and Stillness: The Tale of Myotonia in an Adult Warrior: A Case Report

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Abstract

Myotonic Dystrophy type 1 (DM1) is an autosomal dominant multisystem disorder characterized by progressive muscle weakness, myotonia, and involvement of multiple organ systems, with variable clinical presentations. We present the case of a 39-year-old male soldier who initially sought evaluation for non-anginal chest pain and was subsequently found to have distal upper limb weakness and difficulty making a fist over a two-year period. Neurological examination revealed subtle bilateral lower motor neuron facial palsy, neck flexor weakness, muscle wasting in the left deltoid and supraspinatus, along with percussion and hand grip myotonia. Electromyography showed typical myotonic discharges, and genetic testing confirmed an expanded CTG trinucleotide repeat in the DMPK gene, establishing the diagnosis of DM1. Comprehensive systemic and cardiac evaluation showed no abnormalities. This case highlights the diagnostic challenges posed by the heterogenous manifestations of DM1 and underscores the need for heightened clinical suspicion when encountering patients with subtle neuromuscular symptoms and myotonia. Early identification is essential for appropriate multidisciplinary management that addresses potential complications such as cardiac conduction defects and ultimately improves patient outcomes. Additionally, this report adds valuable insight into the phenotypic spectrum of DM1 in adult patients, emphasizing the importance of detailed clinical and electrophysiological assessment supported by genetic confirmation. Raising awareness about this rare but impactful condition can facilitate timely diagnosis and personalized care.

Keywords: Myotonic Dystrophy type 1; Autosomal dominant disorder; Myotonia; Multisystem involvement; Electromyography; Genetic testing.

1. Introduction

Myotonic dystrophies are the most prevalent muscular dystrophies affecting adults, inherited in an autosomal dominant manner, and primarily characterized by muscle weakness and delayed muscle relaxation (myotonia), along with involvement of multiple body systems. There are two recognized types: Myotonic Dystrophy type 1 (DM1) and type 2 (DM2). Both types share core clinical features such as muscle weakness, myotonia, and early-onset cataracts; however, there exist key differences that help distinguish between them.

These differences include variations in symptom severity, age of onset, muscles primarily affected, and some associated systemic symptoms. DM1 typically has earlier onset and may present in congenital forms, whereas DM2 usually manifests later with generally milder symptoms but prominent proximal muscle involvement. Despite their clinical similarities, the distinct features of DM1 and DM2 allow their differentiation in clinical practice [1,2].

Myotonic Dystrophy type 1 (DM1) is among the most prevalent rare disorders worldwide, with an estimated prevalence of 9.27 cases per 100,000 individuals according to a recent meta-analysis by Liao et al. This analysis included original

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research articles published in English and indexed in PubMed, MEDLINE, Web of Science, and the Cochrane Library up to 2022 [3]. In contrast, Myotonic Dystrophy type 2 (DM2), also known as proximal myotonic myopathy (PROMM), shares clinical features with DM1 but typically manifests in adulthood and with a less severe clinical presentation. Notably, no congenital form of DM2 has been reported to date [1].

Both DM1 and DM2 are inherited in an autosomal dominant manner: each child of an individual with a CTG or CCTG repeat expansion has a 50% chance of inheriting the expansion. For DM2, de novo pathogenic variants have not been reported to date: all affected individuals have had one parent with a CCTG repeat expansion.

2. Case report

2.1. Presentation

A 39 years old male, a chronic smoker, occasionally consuming alcohol, presented with non-anginal chest pain. Further history taking revealed difficulty in making fists for 02 years, observed first when he was unable to get a grip of a rope while doing Vertical Rope Climb (exercises). He sought medical attention elsewhere in view of persistent symptoms. By this time, he gradually gained weight. He was found to have Hypothyroidism (TSH-13mIU/L) and was initiated on medications. However, the symptoms persisted. The non anginal chest pain made the patient to seek medical attention again.

2.2. Examination

General examination including Blood Pressure recordings, Pulse and respiratory rate was within normal limits. General Appearance – Frontal balding was present (Figure 1).

Figure 1 Frontal Balding in the patient

Figure 2 Wasting of left deltoid and supraspinatus

Cardiological examination was normal. Neurological examination revealed bilateral LMN type facial palsy, weakness in neck flexion and wasting of left deltoid and supraspinatus muscle (Figure 2).

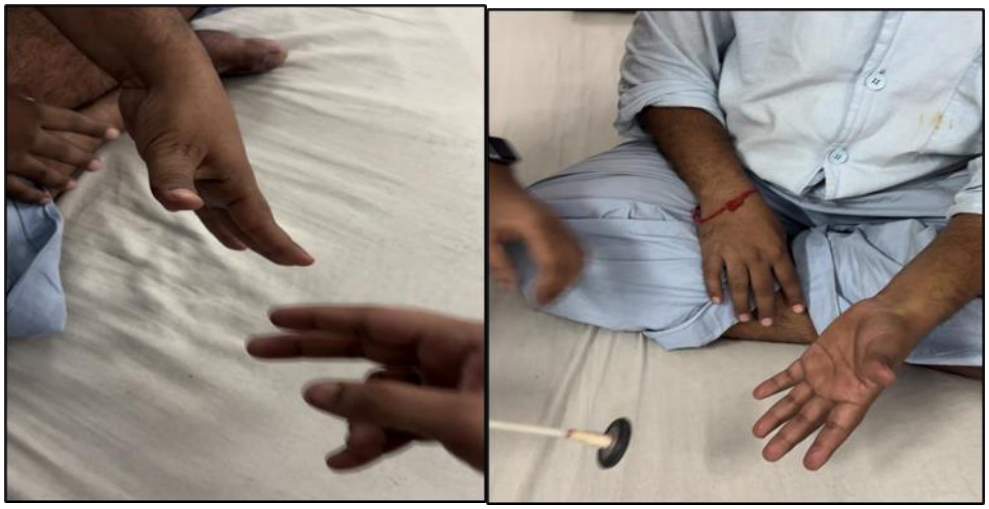

Figure 3 Hand grip myotonia and Percussion myotonia being demonstrated

Power in Bilateral Upper limb abduction was 4. Deep Tendon Reflexes 1+ over Biceps, 1+ over Supinator, 1+ over Triceps, 2+ over Knee and Absent Ankle Jerks. Percussion myotonia and hand grip myotonia was present (Figure 3).

2.3. Investigation

His Biochemical parameters including CKMB were normal. MRI Brain with cervical spine revealed normal study. Electromyography (EMG) findings are suggestive of myotonic discharges in the bilateral abductor pollicis brevis (APB), first dorsal interosseous (FDI), and left deltoid muscles. Genetic analysis (WES for myotonic Dystrophy) confirmed DMPK CTG (Dystrophia Myotonica Protein Kinase Cytosine-Thymine-Guanine trinucleotide repeat sequence) repeats > 50. Further systemic evaluation revealed no cardiac conduction abnormalities, no cataracts, no features of IBS/dysphagia/gall stones with normal HBA1C levels.

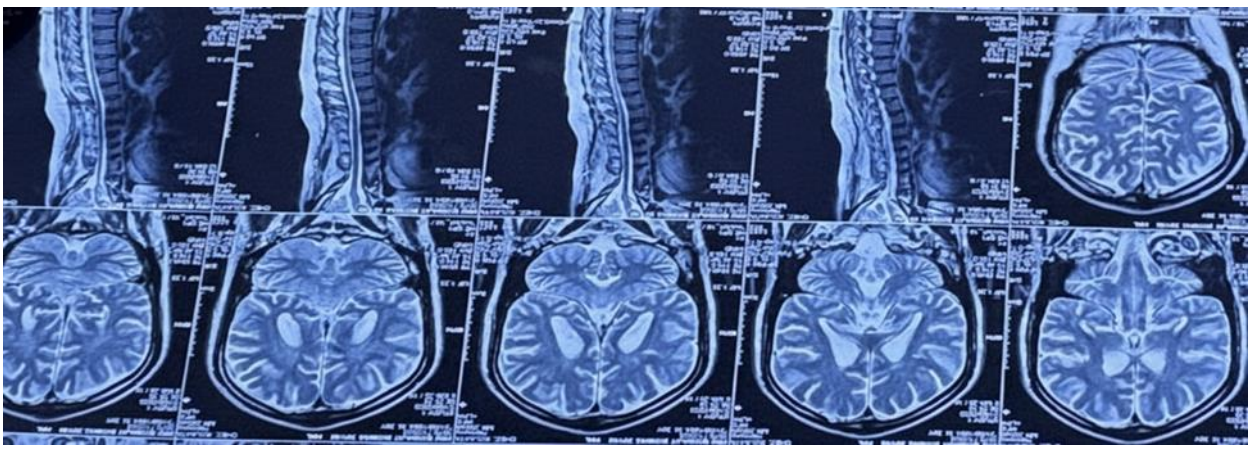

Figure 4 Normal Magnetic Resonance Imaging of Brain and Spinal Cord

The patient received supportive treatment including genetic counseling and symptomatic management aimed at preserving functional independence. A multidisciplinary supportive approach focused on symptom relief, functional preservation, and monitoring for complications, aiming to optimize quality of life through coordinated care across relevant specialties.

3. Discussion

Myotonic dystrophies attract significant research attention due to their clinical significance and fascinating molecular mechanisms. The primary features of myotonic dystrophy type 1 (also called Steinert's disease) and type 2 include progressive muscle degeneration causing disabling weakness and wasting, accompanied by myotonia and involvement of multiple body systems [4,5].

Patients with myotonic dystrophy type 1 (DM1) may initially present with one of four forms: adult-onset, congenital, childhood-onset, or late-onset with few symptoms (oligosymptomatic). Among these, adult-onset DM1 is the most frequently encountered form [4].

Diagnostic evaluations for myotonic dystrophy type 1 are often prompted by the presence of the three primary symptoms: muscle weakness, myotonia, or cataracts. Additionally, a family history of type 1 myotonic dystrophy along with milder symptoms frequently serves as a basis for initiating diagnostic investigations. In our patient, the muscle weakness was an additional complaint and careful and diligent clinical examination led to further evaluation and clinching of diagnosis.

Myotonia is often described as muscle stiffness by patients or their parents, typically noticed from school age through the third decade of life, or may be discovered later during clinical or electrophysiological evaluations. In early adulthood, muscle weakness may be completely absent. Cataracts, which often prompt ophthalmologic evaluation and surgical intervention, usually do not lead to consideration of myotonic dystrophy type 1 as a diagnosis, especially when detected in older individuals.

In adult-onset myotonic dystrophy type 1, progressive skeletal muscle weakness results in severe disability and is a leading cause of death in the later stages. This weakness primarily affects the facial, neck, and distal limb muscles, accompanied by muscle wasting. The characteristic myopathic facial appearance, marked by temporal muscle atrophy and drooping eyelids (ptosis), is often accentuated by frontal hair thinning in men. Myotonia is consistently detected through clinical examination and electromyography, though it may sometimes be subtle. The most common clinical sign is percussion-induced myotonia in the thenar muscles, with grip myotonia occurring less frequently. Repeated muscle activation typically alleviates myotonia, known as the warm-up phenomenon [7].

Cardiac conduction abnormalities and rapid arrhythmias can result in early episodes of cardiac dysfunction and sudden cardiac death. The primary pathological change involves fibrosis within the conduction pathways and the sinoatrial node. Although subclinical cardiac changes may be detected, overt dilated or hypertrophic cardiomyopathy causing heart failure is not consistently observed in adult-onset myotonic dystrophy type 1 [8].

Cataracts in adult-onset myotonic dystrophy type 1 typically present as iridescent opacities in the posterior subcapsular region of the lens and are considered nearly pathognomonic for the condition [9]. Often, the precise cataract type is not identified prior to surgical intervention. Clinicians should suspect myotonic dystrophy when cataracts develop before the age of 50.

Individuals with diseases caused by DNA repeat expansions exhibit longer and unstable repeat sequences compared to unaffected individuals. The length of these repeats varies across generations, within an individual's lifespan, among different tissues of the same person, and even within a single tissue over time. This instability occurs in both dividing (mitotic) and non-dividing (post-mitotic) cells, with disease manifestations being particularly prominent in post-mitotic tissues such as the brain, heart, and skeletal muscle. Pathological expansions lead to the formation of abnormal DNA, DNA-RNA, and RNA structures that disrupt nucleic acid processes including replication, repair, recombination, and transcription [10].

The diagnosis of DM is commonly made based on clinical presentation coupled with a positive family history. Confirmation of DM1 diagnosis is achieved through genetic testing, specifically identifying an expanded CTG repeat in the DMPK gene, which is the definitive standard. If myotonia is not clinically evident or diagnosis remains unclear, electromyography (EMG) typically reveals its presence. Among the three recognized phenotypes of myotonic dystrophy type 1, this particular case falls under the classical category based on the estimated CTG repeat size.

At present, there is no officially approved curative therapy for patients with DM. Treatment is primarily supportive, focusing on careful monitoring for the development of disease-related complications [1]. Management approaches for DM2 closely resemble those used for DM1, as few treatments are specifically designed for DM2. Key components in managing DM patients include genetic counseling, efforts to preserve functional independence, addressing the

multisystemic aspects of the disease, and relieving symptoms. Although referral to and ongoing monitoring by a neuromuscular specialist are critical, the complexity of DM requires a multidisciplinary care team. Coordination with cardiologists, pulmonologists, ophthalmologists, physiatrists, gastroenterologists, endocrinologists, and geneticists is essential to deliver comprehensive and optimal care. Effectively managing treatable multisystem complications and symptoms not only improves quality of life but also may prolong survival.

Abbreviations

- DM1: Myotonic Dystrophy type 1
- DM2: Myotonic Dystrophy type 2
- PROMM: Proximal Myotonic Myopathy
- TSH: Thyroid Stimulating Hormone
- LMN: Lower Motor Neuron
- CKMB: Creatine Kinase-MB
- MRI: Magnetic Resonance Imaging
- EMG: Electromyography
- APB: Abductor Pollicis Brevis
- FDI: First Dorsal Interosseous
- WES: Whole Exome Sequencing
- DMPK: Dystrophia Myotonica Protein Kinase
- CTG: Cytosine-Thymine-Guanine (trinucleotide repeat)
- IBS: Irritable Bowel Syndrome
- HBA1C: Glycated Hemoglobin A1c

4. Conclusion

This case highlights the diagnostic challenges and multisystemic nature of Myotonic Dystrophy type 1 (DM1), emphasizing the importance of thorough clinical evaluation in patients presenting with subtle neuromuscular symptoms and non-specific complaints such as non-anginal chest pain. Early recognition through detailed neurological examination, electrophysiological studies, and confirmatory genetic testing is critical for timely diagnosis and appropriate management. Given the variable clinical phenotype and potential involvement of multiple organ systems, a multidisciplinary approach is essential to optimize patient care and improve quality of life. This case further underscores the need for increased awareness among clinicians to consider DM1 in differential diagnoses, especially in adult patients with unexplained muscle weakness, myotonia, and systemic features. Continued research and genetic counseling remain pivotal components in the management of affected individuals and their families.

Compliance with ethical standards

Disclosure of conflict of interest

No conflicts of Interest. In accordance with the ICMJE uniform disclosure requirements, the authors declare the following:

- *Payment/services*

No financial support was received from any organization for the submitted work.

- *Financial relationships*

The authors report no financial relationships, either current or within the past three years, with organizations that could have an interest in this work.

- *Other relationships*

The authors also declare no other relationships or activities that could be perceived as influencing the submitted work.

Statement of Ethical Approval

Ethical approval was not applicable for this case report as it does not constitute a research study. Patient informed consent for publication of clinical details and images was obtained.

Statement of informed consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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