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A case report: Adult-onset Myotonic Dystrophy Type 1 Mimicking Type 2 with Predominant Proximal Weakness

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INTRODUCTION

Myotonic dystrophy (DM) is an autosomal dominant multisystem disorder. DM type 1 (DM1) is caused by CTG repeat expansion in the DMPK gene and is classically associated with myotonia and distal-predominant weakness, whereas DM type 2 (DM2) more frequently presents with adult-onset proximal weakness. However, DM1 can show considerable phenotypic heterogeneity, which may lead to subtype misclassification based on clinical features alone.

CASE

A 54-year-old man presented with a 10-year history of progressive stooped posture and gradually worsening weakness in both upper and lower extremities. The weakness pattern was predominantly proximal/axial with a relatively mild course, initially suggesting DM2. He had a family history of muscle disease (father and grandaunt).

Physical examination revealed mild percussion myotonia and grip myotonia, and mild frontal balding with bitemporal recession. Manual muscle testing demonstrated proximal-predominant weakness, most marked in the shoulder girdle and hip extensors, with relative preservation of finger flexion and knee extension. Laboratory findings were unremarkable except for a mild elevation in serum creatine kinase (384 U/L).

Nerve conduction studies were within normal limits. Needle electromyography showed widespread myotonic discharges with wax-and-waning amplitude and frequency in most tested muscles, with positive sharp waves and fibrillation potentials and short-duration polyphasic motor unit potentials; maximal effort recruitment ranged from partial to complete interference patterns (PIP–CIP). Overall electrophysiologic findings were supportive of myotonic dystrophy. Genetic testing identified a pathogenic DMPK CTG repeat expansion in the full mutation range (reported as >90 repeats in one allele), confirming DM1 despite the DM2-like weakness distribution.

Table 1		Right	Left
Shoulder	Flexion	P+	F-
	Extension	P+	P+
Elbow	Flexion	F	F+
	Extension	F	F
Wrist	Flexion	F+	F+
	Extension	F+	F+
Finger	Flexion	G	G
Hip	Flexion	F+	F+
	Extension	F-	F-
Knee	Flexion	F+	F+
	Extension	G	G
Ankle	Dorsi flexion	F+	F+
	Plantar flexion	F+	F+

Figure 1. Patient's photograph demonstrating frontal balding with bitemporal recession (M-shaped hairline)
Figure 2. Percussion was applied over the right thenar eminence (A), resulting in sustained contraction of the thenar muscles with delayed relaxation (B), an examination finding consistent with mild percussion myotonia.

Table 1. Manual muscle testing demonstrating predominantly proximal weakness involving both upper and lower extremities.
※ Abbreviations : (N, normal/grade 5; G, good/grade 4; F, fair/grade 3; P, poor/grade 2; T, trace/grade 1; Z, zero/grade 0)

CONCLUSION

This case highlights that DM1 may present as an adult-onset, mild phenotype with predominant proximal/axial weakness and stooped posture, mimicking DM2. The presence of subtle myotonia, frontal balding, and an autosomal dominant family history should prompt consideration of DM1 even when distal weakness is not prominent. Molecular confirmation is essential when clinical subtype features overlap.