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Co-occurrence of myotonic dystrophy type 1 and hereditary neuropathy with liability to pressure palsies: A case report

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Abstract

This report is the first documented co-occurrence of myotonic dystrophy type 1 (DM1) and hereditary neuropathy with liability to pressure palsies (HNPP); rare neuromuscular disorders with distinct genetic etiologies (DMPK CTG expansion, PMP22 deletion). A 22-year-old soldier underwent clinical evaluation for weakness of both fingers after physical training. Clinical features included percussion myotonia, grip difficulty (DM1), and limb numbness, weakness, absent reflexes (HNPP). NCS showed widespread demyelination; EMG confirmed myotonic discharges. This novel overlap underscores the need for comprehensive neurophysiological and genetic evaluations in atypical presentations. The absence of prior reports suggests coincidental co-occurrence, emphasizing vigilance for multiple pathologies.

Keywords: Myotonic dystrophy type 1 (DM1), hereditary neuropathy with liability to pressure palsies (HNPP), case reports, phenotype

INTRODUCTION

Myotonic dystrophy type 1 (DM1) is an autosomal dominant myopathy that frequently affects skeletal muscle, the heart, eyes, and the central nervous system. It is primarily caused by a CTG trinucleotide repeat expansion in the dystrophia myotonica protein kinase (DMPK) gene on chromosome 19. A higher number of repeats is generally associated with an earlier age of onset and a poorer prognosis.[1,2] Hereditary neuropathy with liability to pressure palsies (HNPP) results from a deletion of the peripheral myelin protein 22 (PMP22) gene on chromosome 17, leading to myelin sheath instability and focal nerve palsies upon minor mechanical compression, with a peak incidence in young adulthood.[3,4] DM1 has an estimated prevalence of 1/8 000 worldwide[1], HNPP occurs in 16/100 000 individuals.[2] The co-occurrence of both disorders in a single individual is therefore exceedingly rare.

CASE REPORT

We report the case of a 22-year-old male soldier with a previously unremarkable medical history, who is not married and has no children. In July 2024, following horizontal bar training, he developed weakness in bilateral finger flexion and difficulty extending his fingers after making a fist. Subsequently, he experienced numbness and weakness in all four limbs, with distal symptoms being more severe than proximal ones. An electromyography (EMG) performed at another hospital suggested bilateral median nerve injury, and his symptoms partially resolved after neurotrophic therapy with vitamin B1 and mecobalamin. Upon admission to our hospital in October, physical examination revealed percussion myotonia of the tongue. He had difficulty relaxing his grip after clenching his fists and exhibited reduced distal muscle strength (grade 4), consistent with the diagnosis of DM1. Additionally, tendon reflexes in all limbs were diminished or absent, and hypoalgesia was noted

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below the right wrist, indicative of a peripheral neuropathy.

Laboratory tests showed elevated creatine kinase (CK) (298 U/L; reference range, 30–200 U/L) and creatine kinase-MB (CK-MB) (5.9 μg/L; reference range, 0–5.2 μg/L), suggesting mild myocardial injury. Cerebrospinal fluid analysis revealed increased levels of albumin (443 mg/L; reference range, 0–350 mg/L), IgA (7.35 ng/L; reference range, 0–5 ng/L), and IgG (53.4 ng/L; reference range, 0–34 ng/L), with a few lymphocytes, indicating altered permeability of the blood-brain barrier. An electrocardiogram demonstrated sinus bradycardia, a prolonged PR interval, and left axis deviation, suggestive of cardiac involvement. Magnetic resonance imaging (MRI) of the brain and quadriceps muscles showed no abnormalities.

Nerve conduction studies (NCS) (Figure 1A–C) revealed widespread demyelinating damage. The distal motor latencies of bilateral median and ulnar nerves were prolonged, with reduced conduction velocities and decreased amplitudes. A conduction block was identified at the elbow segment of the ulnar nerve. Sensory nerve conduction was also abnormal, with no sensory nerve action potential elicited from the right median nerve. The lower limb nerves showed subclinical demyelinating changes, consistent with the features of HNPP.[3] Electromyography (EMG) (Figure 1D, E) showed short-duration, low-amplitude, polyphasic motor unit potentials accompanied by prominent myotonic discharges, which are characteristic of DM1.[1]

A biopsy of the left biceps brachii muscle revealed no typical myopathic changes (Figure 2A).

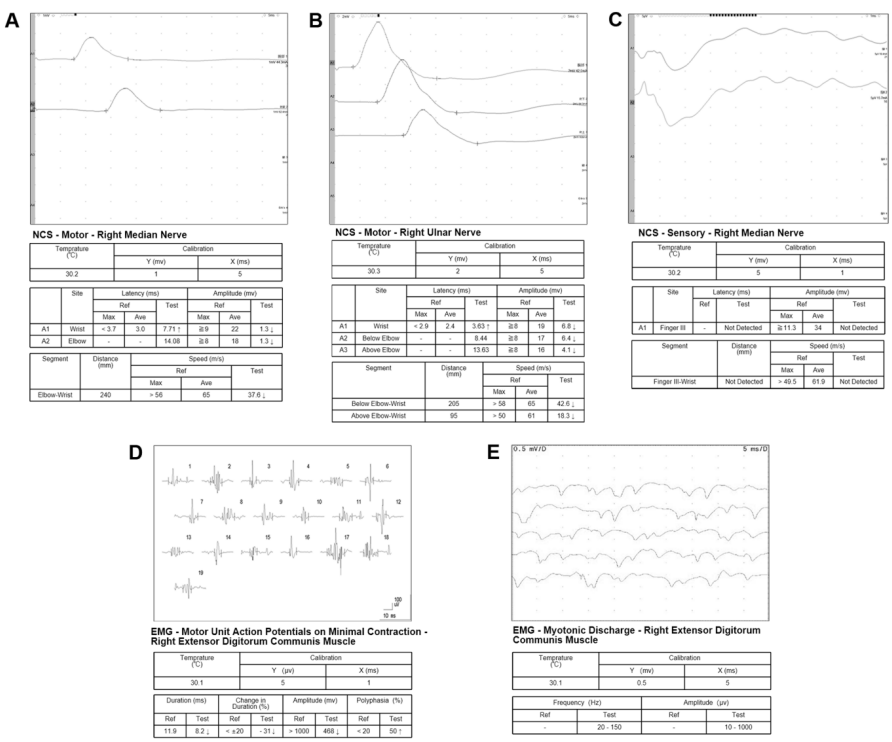

Figure 1. Neurophysiological examination results

(**A**)Right median motor nerve conduction: prolonged distal latency and decreased amplitude at the wrist; decreased amplitude at the elbow; reduced conduction velocity from elbow to wrist. (**B**) Right ulnar motor nerve conduction: prolonged distal latency and decreased amplitude at the wrist; conduction block between the above-elbow and below-elbow sites with reduced conduction velocity; decreased amplitude and reduced conduction velocity from below elbow to wrist. (**C**) Right median sensory nerve conduction (recording from digit III): no reproducible waveform was elicited. (**D**) Right extensor digitorum communis muscle: myogenic pattern with motor unit potentials characterized by short duration, low amplitude, and increased polyphasic waves. (E) Right extensor digitorum communis muscle: abundant myotonic discharges were observed, appearing as runs of positive sharp waves or fibrillation-like potentials with a waxing and waning pattern in both amplitude and frequency, producing a sound reminiscent of a motorcycle engine. The discharge duration lasted approximately 50 ms.

Genetic analysis identified two alleles for the DMPK gene with 12 and >200 CTG repeats, respectively (pathogenic threshold >50 repeats) (Figure 2B, C), confirming the diagnosis of DM1.[1] The analysis also detected a heterozygous deletion of ~ 1.38 Mb in the 17p11.2 region (chr17:14,095,306-15,472,344), which includes the whole PMP22 gene, confirming the diagnosis of HNPP.[3] (Figure 2D).

During hospitalization, the patient received neurotrophic therapy (vitamin B1, 100 mg, IM, qd; mecobalamin, 0.5 mg, IV, qd) and rehabilitation treatments (acupuncture, low-frequency pulsed electrical stimulation, magnetothermal therapy, and transcranial magnetic stimulation, each once daily), resulting in subjective symptomatic improvement.

Prior to discharge, the patient received the following counseling and recommendations:

- Attend annual follow-up appointments to monitor for disease progression, including assessments of muscular, neurological, cardiac, respiratory, endocrine, reproductive, and ocular systems. Seek prompt medical evaluation for any new or worsening symptoms.
- Avoid postures and activities that compress

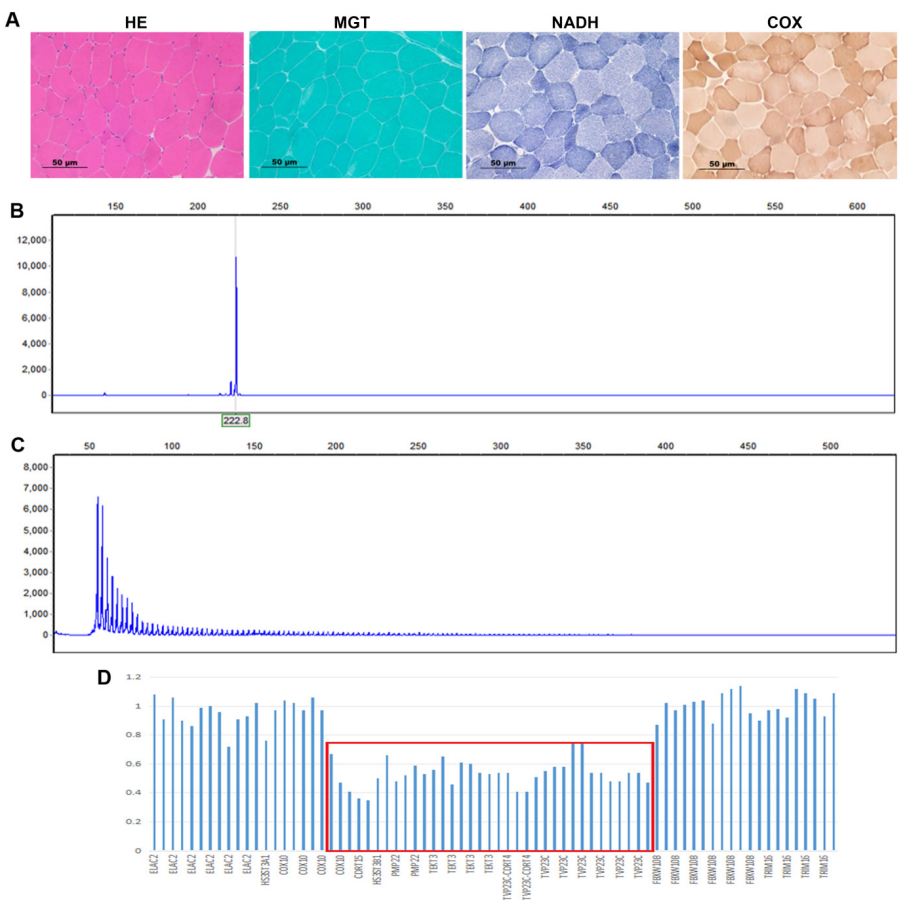

Figure 2. Muscle biopsy and genetic testing results

(**A**) Frozen sections of the left biceps brachii muscle (400×, scale bar = 50 μm). HE showed normal morphology; MGT revealed no ragged red fibers; NADH-TR showed normal distribution of fiber types; COX showed normal enzyme activity. These findings highlight the limitations of muscle biopsy for diagnosing certain hereditary myopathies, particularly in early disease stages or when sampling specific muscles. (**B**) DMPK gene PCR followed by capillary electrophoresis revealed an abnormal expanded peak, corresponding to 12 and >200 CTG repeats on the two alleles. (**C**) DMPK gene triplet-primed PCR (TP-PCR) with capillary electrophoresis displayed the characteristic staircase pattern of peaks. Results from B and C support the diagnosis of DM1. (**D**) Whole-exome target-capture sequencing of the 17p12-p11.2 region (chr17:14,095,306-15,472,344) identified a heterozygous ~1.38 Mb deletion encompassing the PMP22 locus. These findings support the diagnosis of HNPP.

peripheral nerves—such as crossing legs, leaning on elbows, or resting the head on an arm—to reduce the risk of HNPP-related palsy.

- Disclose the full diagnosis of both DM1 and HNPP to any medical team prior to surgeries or procedures, due to increased sensitivity to anesthetics and muscle relaxants in DM1 and elevated risk of compression neuropathy in HNPP.
- Pursue genetic counseling before family planning, given the autosomal dominant inheritance of both conditions. Be aware that DM1 exhibits anticipation, with potential for earlier onset and more severe symptoms in offspring.
- Consider transitioning from military service to an occupation with minimal physical demands to avoid mechanical stress that may trigger HNPP symptoms.
- Avoid high-intensity independent exercise; participate only in professionally supervised rehabilitation and physical training programs.
- Seek psychological counseling or support services to address any emotional distress or adjustment difficulties related to the diagnosis.

At a follow-up appointment in September 2025, the results of his electrocardiogram, EMG, and serological tests were not significantly changed from baseline. Muscle strength in the limbs had recovered to grade 5, although grip strength and dorsiflexion strength remained at grade 4. The mild hypoalgesia below his right wrist persisted without progressive worsening.

DISCUSSION

To our knowledge, this is the first reported case of the co-occurrence of DM1 and HNPP. The clinical presentation represented an overlap of myotonia characteristic of DM1 and demyelinating peripheral neuropathy characteristic of HNPP. The muscle biopsy did not detect pathological changes in the early stage of this patient's disease. Highlighting the diagnostic reliance on the combination of neurophysiological studies and genetic testing. Currently, there are no curative treatments for either condition; management focuses on symptomatic control and the prevention of complications.[1,3]

A previous report by Hodapp *et al*.[5] described a female patient with early-onset disease who had both a DMPK CTG expansion (>1000 repeats) and a PMP22 duplication. Her disease began at age 4, progressed steadily, and she died of respiratory failure at age 32. The PMP22 abnormality was inherited from her mother, while the DMPK mutation was presumed to be paternal. The patient in our case reported that his parents and sister were asymptomatic. Unfortunately, the family declined to participate in a genetic investigation, limiting further clarification of the inheritance pattern.

One year after onset, the patient's condition is stable. However, whether the co-occurrence of DM1 and HNPP influences their respective natural histories remains unknown. For instance, it is unclear whether the muscle atrophy associated with DM1 might increase the susceptibility of nerves to compression or impair their regenerative capacity. Therefore, long-term, regular, and multisystemic follow-up is crucial. This rare case presents diagnostic challenges, complexities in management, and has profound genetic implications. It underscores the importance of maintaining an open diagnostic mindset when faced with complex clinical presentations, utilizing advanced diagnostic technologies for precise diagnosis, and formulating highly individualized management strategies to ultimately improve the patient's long-term prognosis and quality of life.

DISCLOSURE

Ethics: This case report was approved by the Research Ethics Committee of the 960th Hospital of PLA (No. 2025-181). Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Data availability: The data supporting the findings of this case report are available from the corresponding author upon reasonable request.

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Conflict of interest: None

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