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# A Coincidental Existence of Myotonic Dystrophy and Charcot-Marie-Tooth disease: A Case report

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**Introduction**

Myotonic dystrophy type 1 (DM1) is an autosomal dominant multisystem disorder and one of the most common muscular dystrophies affecting adults. Charcot-Marie-Tooth (CMT) disease, a common hereditary neuropathy, is characterized by atrophy of the distal limbs and peripheral nerve abnormalities. The authors report a rare case involving a 24-year-old female who was diagnosed simultaneously with both DM1 and CMT1A based on the Results of a nerve conduction study (NCS).

**Case report**

24-year-old female, with complaints of bilateral lower extremity weakness and an inability to run since she was 10 years of age, was admitted for bilateral lower extremity pain in 2010. No other medical history, such as diabetes mellitus, was reported. On physical examination, the patient exhibited signs indicative of percussion myotonia, grip-release myotonia, and cramping. In addition, she also exhibited atrophy in both calf muscles and bilateral high arched foot. A manual muscle test revealed a good grade in all extremities, but only a fair grade in the ankle joint. No spasticity was noticed, and decreased deep tendon reflexes of all extremities were identified. She reported a tingling sensation below the knee and in both upper extremities. An EDX was performed to investigate weakness in all extremities and paresthesia. A motor and sensory NCS of all extremities revealed abnormalities. F-wave for the upper and lower extremities and H-reflex showed no response. Needling EMG revealed increased insertional activity, abnormal spontaneous activity, including positive sharp waves, and myotonic discharges in both the right upper and lower extremities. Based on physical examination and EDX abnormalities, the patient had a high probability of being diagnosed with myotonic dystrophy combined with chronic inflammatory demyelinating polyneuropathy. Genetic evaluation (DMPK gene study) was performed and revealed an abnormal number of CTG repeats that confirmed classic DM type 1. Five years later, the patient was admitted for dizziness with headache and lower extremity pain. Among all evaluations, EDX findings continued to reveal NCS abnormalities, indicating sensorimotor demyelinating polyneuropathy. Therefore, an additional genetic study was performed to identify whether she had other hereditary neuropathies in addition to DM1. Peripheral myelin protein-22, PMP-22 for CMT1A and myelin protein zero, MPZ gene for CMT1B were examined, and a novel diagnosis of CMT1A was made by validating the duplication of the PMP-22 gene.

**Discussion**

The present case demonstrates that DM1 and CMT, which are different hereditary neuromuscular disorders, can be diagnosed at the same time, and that EDX is an essential element in determining the comorbid condition. Severe serial abnormalities in an NCS in a DM1 patient may suggest the incidental coexistence of hereditary neuropathies, and further evaluations, such as genetic studies, should be performed for proper diagnosis.