**Charcot-Marie-Tooth Disease, So Which Type?**

Ayşe Nur Coşkun1, Canan Üstün1, Özgen Hür1, Deniz Torun2, Mutluay Arslan1, Bülent Ünay1

1 University of Health Sciences, Gulhane Faculty of Medicine, Department of Pediatric Neurology, Turkey

2 University of Health Sciences, Gulhane Faculty of Medicine, Department of Medical Genetics, Turkey

Objectives: Autosomal recessive demyelinating Charcot-Marie-Tooth disease is a phenotypic and genetically heterogeneous disease. This article aimed to emphasize type 4C Charcot-Marie-Tooth disease due to SH3TC2 gene mutation.

Case: A ten-year-old male patient was evaluated for scoliosis and pes cavus. The patient, who had kyphoscoliosis, had facial and neck muscle weakness, tongue atrophy, bilateral pectoral and thenar-hypothenar atrophy, decreased bilateral lower extremity muscle strength (4/5) and bilateral pes cavus in his neurological examination. Bilateral lower extremities deep tendon reflexes were absent. The electromyographic study of the patient was consistent with severe sensorimotor demyelinating neuropathy. Cranial magnetic resonance imaging and echocardiographic findings were normal. There was consanguinity between mother and father. The patient's PMP22 gene analysis was normal. In the whole exome sequence analysis of the patient, c.3328\_3329insGCTTCTGTTCCTAGG homozygous pathogenic mutation was detected in the SH3TC2 gene. This variant has not been reported before, but evaluations in databases show the variant as likely pathogenic. It was confirmed by Sanger sequence analysis, and it was seen that both parents were heterozygous carriers for this mutation. The patient was diagnosed with Charcot-Marie-Tooth disease type 4C.

Conclusion: The common genetic cause of Charcot-Marie-Tooth disease is the PMP22 gene mutation. Therefore, a single-gene analysis should be performed regarding PMP22 deletion or duplication. In patients with normal single gene analysis, whole exome or whole genome sequence analysis should be performed.

Keywords: "pes cavus, scoliosis, severe sensorimotor demyelinating neuropathy."