Familiar hypokalemic periodic paralysis: An uncommon cause of acute flaccid paralysis

Joana Aquino1, Margarida Henriques1, Ana Paula Rocha1, Ana Luisa Reis1, Margarida Pereira1 and Sergio Sousa2
Centro Hospitalar de Leiria, Portugal
Centro Hospitalar e Universitario de Coimbra, Portugal

Introduction: Familiar hypokalemic periodic paralysis is an autosomal dominant transmission channelopathy characterized by

recurrent episodes of reversible muscle weakness and hypokalemia.

Case Study: We describe the case of a 16 year-old boy with recurrent episodes of decreased muscle strength whose mother had a history of similar episodes. He was admitted in the emergency department with flaccid tetraparesis and hypokalemia (1.4 mmol/L) which resolved after potassium administration. CACNA1S gene molecular study identified the mutation c.1583G>A-(p.Arg528His). Currently he is under an optimized nutrition plan, avoidance of triggers and acetazolamide and potassium chloride therapeutic.

Discussion: Although rare, familiar hypokalemic periodic paralysis' timely recognition is crucial to initiate proper treatment and avoid associated complications. In this case, genetic testing confirmed the diagnosis and allowed the identification of asymptomatic relatives at risk of sudden acute paralysis and anesthetic complications.