

Charcot-Marie-Tooth Disease: Case Report of An Uncommon Cause of Lower Limb Weakness in Malaysia

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Charcot-Marie-Tooth (CMT) disease is an inherited neuropathy that encompasses a genetically heterogeneous group of disorders affecting the peripheral nervous system, despite typically manifesting as a relatively homogeneous clinical phenotype characterised by progressive distal muscle weakness, atrophy, foot deformities and sensory deficits. We aim to highlight CMT as an uncommon aetiology of lower limb weakness in Malaysia and discuss its diagnostic challenges. A 53-year-old woman with no underlying illnesses reported a two-year history of progressive bilateral lower limb weakness originating in distal muscles. She required a walking stick due to deteriorating gait, but otherwise denied sensory or autonomic symptoms. The patient had consulted multiple general practitioners, who primarily diagnosed her with vitamin B deficiency without detailed examinations. However, her symptoms failed to resolve with vitamin B complex. On examination, symmetrical peroneal muscle wasting was observed in lower extremities, resulting in a classical inverted champagne bottle appearance. Motor power was 4/5 for bilateral dorsiflexion and plantarflexion, while proximal muscles retained full power. Reflexes were 1+ in lower limbs and 2+ in upper limbs. Additional findings included pes cavus, impaired proprioception and positive Romberg's test. These raised suspicions of CMT, leading to referral to a neurology team for further diagnostic evaluations. CMT is a complex disorder with various subtypes, each exhibiting distinct clinical features and genetic mutations. Limited awareness among healthcare professionals often leads to misdiagnosis. We suggest future research to expand on developing an algorithmic approach that aids in early recognition of CMT as a potential cause for lower limb weakness.

Keywords: Charcot-Marie-Tooth, lower limb weakness, pes cavus

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