A CHARCOT-MARIE-TOOTH NEUROPÁTIA KLINIKAI, GENETIKAI KÉPE ÉS A BETEGEK REHABILITÁCIÓJA

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Abstract: Charcot-Marie-Tooth disease (CMT) represents a group of clinical and genetical heterogenous disorders affecting the peripheral nervous system. CMT is clinically characterized by progressive weakness and atrophy of distal muscles of both lower and upper extremities. Neurophysiological, as well as histopathologic criteria differentiates CMT disease into: CMT type 1 (CMT 1) or hereditary motor and sensory neuropathy type 1 (HSMN I), CMT type 2 (CMT 2) or HSMN II, the neuronal form of CMT because of axonal degeneration of peripheral nerves. Most of CMT patients belong to families in which the disease segregates according to an autosomal dominant inheritance pattern. There are reports of families with an X-linked or recessive mode of inheritance as well as sporadic patients. The goals of rehabilitation in patients with CMT disease are to maximize and prolong independent function and locomotion, and provide acces to full integration into society.

Keywords: Charcot-Marie-Tooth disease, clinical aspects, genetical aspects, rehabilitation