Peripheral Nerve Disorders: Chapter 47. Dominant Charcot-Marie-Tooth syndrome and cognate disorders (Handbook of Clinical Neurology)

Davide Pareyson, Chiara Marchesi, Ettore Salsano

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Peripheral Nerve Disorders: Chapter 47. Dominant Charcot-Marie-Tooth syndrome and cognate disorders (Handbook of Clinical Neurology) Davide Pareyson, Chiara Marchesi, Ettore Salsano Charcot–Marie–Tooth neuropathy (CMT) is a group of genetically heterogeneous disorders sharing a similar phenotype, characterized by wasting and weakness mainly involving the distal muscles of lower and upper limbs, variably associated with distal sensory loss and skeletal deformities. This chapter deals with dominantly transmitted CMT and related disorders, namely hereditary neuropathy with liability to pressure palsies (HNPP) and hereditary neuralgic amyotrophy (HNA). During the last 20 years, several genes have been uncovered associated with CMT and our understanding of the underlying molecular mechanisms has greatly improved. Consequently, a precise genetic diagnosis is now possible in the majority of cases, thus allowing proper genetic counseling. Although, unfortunately, treatment is still unavailable for all types of CMT, several cellular and animal models have been developed and some compounds have proved effective in these models. The first trials with ascorbic acid in CMT type 1A have been completed and, although negative, are providing relevant information on disease course and on how to prepare for future trials.

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