Peripheral Nerve Disorders: Chapter 48. Recessively transmitted predominantly motor neuropathies (Handbook of Clinical Neurology)

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Recessively transmitted predominantly motor neuropathies are rare and show a severe phenotype. They are frequently observed in populations with a high rate of consanguineous marriages. At least 15 genes and six loci have been found to be associated with autosomal recessive CMT (AR-CMT) and X-linked CMT (AR-CMTX) and also distal hereditary motor neuronopathy (AR-dHMN). These disorders are genetically heterogeneous but the clinical phenotype is relatively homogeneous. Distal muscle weakness and atrophy predominating in the lower extremities, diminished or absent deep tendon reflexes, distal sensory loss, and pes cavus are the main clinical features of this disorder with occasional cranial nerve involvement. Although genetic diagnosis of some of subtypes of AR-CMT are now available, rapid advances in the molecular genetics and cell biology show a great complexity. Animal models for the most common subtypes of human AR-CMT disease provide clues for understanding the pathogenesis of CMT and also help to reveal possible treatment strategies of inherited neuropathies. This chapter highlights the clinical features and the recent genetic and biological findings in these disorders based on the current classification.

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