

Abstract

Hereditary motor and sensory neuropathy (in short HMSN or hereditary sensomotoric neuropathy) also known as Charcot-Marie-Tooth disease (CMT) is a clinically and genetically heterogeneous group of diseases which are the most frequent disorders affecting peripheral nervous system. The prevalence of these illnesses is generally 5 - 40 people per 100 000 inhabitants. CMT was first described in the 1886. Because of a large number of various types of mutations classification of HMSN is disunited. The main division of CMT depends on the median motor conduction velocity (and also on the part of nerve that was damaged). It is demyelinating (CMT 1) type and axonal (CMT 2) type. Further classification depends on the mode of heredity and phenotypic expression. Autosomal dominant CMT are divided into four main types – CMT 1A to D. Similar, CMT 2 could be distinguished by genetical subtype as well or, the classification can follow phenotypic expression. Beside the autosomal inherited HMSN, other types of hereditary sensomotoric diseases do exist: intermediate CMT, X-linked CMT, Déjerine-Sottas syndrom, congenital hypomyelination neuropathy and hereditary neuropathy with liability to pressure palsy. Individual types of HMSN are caused by mutations in various genes that are localized on different chromosomes. The most common type of CMT is autosomal dominant type CMT 1A. It is caused by the duplication in the gene *PMP22*.

Even though these diseases are not mortal, they significantly affect patients' life. The first clinical symptoms appear in the 1st or 2nd decade of the patient's life. Gradually during the patient's life there appear symptoms such as weakness of lower and lately upper limbs, muscle atrophy, the deformity (mostly) of the feet, sensory weakness and other symptoms, mainly depending on the type of disease/mutation. Molecular-genetic and EMG methods are used for diagnosis of the disease. Because there is no causal therapy of neurogenetic diseases, the contemporary treatment is directed at alleviation of the symptoms.