Hereditary peripheral neuropathy, known asCharcot Marie Tooth disease (CMT) and with an incidence of 1:2500 -1:10 000, is the most common hereditary neuromuscular disorder. Type CMT 1A is the most common form of CMT refering to the group of primary demyelinitateing motor and sensory peripheral neuropathies. CMT phenotype is clinically characterized by chronic slowly progressive distal muscle weakness and atrophy with hypo or areflexia and mild to moderate acral sensory loss. The lower limbs are predominantly affected.

The aims of this study were to describe the first and most common signs of CMT1A during the first decade of life, to characterize their progression, and evaluate the sensitivity of CMTNS (Charcot-Marie-Tooth neuropath scale) for CMT1A young children. Sixteen children aged 3 to 10 years with genetically proven CMT 1A were examined. All patients were clinically examined, underwent electrophysiological examination, and were scored by CMTNS. Eight were followed for up to two years.

Our data shows that CMT 1A in children under the age of 10 years causes only a mild disability. Initial signs of CMT 1A were difficulty in heel walking (15/16, 93%) and lower limb hypo or areflexia (13/16, 81%). The test of heel walking can be easily used as a screening test for hereditary neuropathies in pediatrics. After a two years follow-up period there was an increase in CMTNS points in 4 patients out of 8. We suggest that CMTNS use may be limited for young children with CMT1A to evaluate the therapeutic effect.

Except for one CMT1A patient, two published atypical phenotype of dHMN (distal hereditary neuropathy) and SPG3A (hereditary spastic paraparesis type 3A) are involved in this study In the first case report there is a Czech family with cranial nerves palsy as an initial feature of a non progressive infantile onset dominant dHMN.