The Charcot-Marie–Tooth (CMT) diseases are the most common inherited neuropathies. CMT is characterized clinically by distal muscle wasting and weakness, reduced reflexes and impaired distal sensation and by a sensory motor neuropathy neurophysiologically. The severity of the disease varies enormously depending to a large extent on the underlying genetic defect. The current clinical classification of CMT is done using electrophysiological criteria into type 1 (demyelinating) and type 2 (axonal) and further sub-classification is done according to inheritance pattern. A solely genetic classsification is not possible at present as all the causative genes for CMT are not known.

Autosomal recessive CMT (AR CMT) forms are rare in European populations. The responsible genes have been discovered just in recent years. The disease has usually early onset and fast progressing and severe course. Mutations in GDAP1 gene (ganglioside- induced differentation associated proteine-1) soon showed to be the most common cause of CMT in families with AR pedigrees. They were found in patients with demyelinating (CMT4A) as well as axonal (CMT4C4) CMT. Common GDAP1 mutations are consquence of founder effect. Mutations in PRX (periaxin) gene are responsible for demyelinating CMT type (CMT4F).

Approximately in a half of the CMT families living in the Czech Republic from a unique CMT database of DNA laboratory of Department of Child Neurology , 2nd School of Medicine, Charles University Prague, the underlying genetic cause after testing for the most common CMT causes remained unknown. The aims of the study was to detect pathogenic mutations in GDAP1 a PRX genes in selected patients from AR CMT families and families with isolated CMT cases, to test prevalent mutations for founder effect and to give detailed clinical and electrophysiological phenotype in patients with the identified mutations.