

My PhD thesis can be divided into two parts:

1. Hereditary motor-sensory neuropathies (HMSN)
2. Selected muscle disorders

The main emphasis was on the first part – hereditary motor and sensory neuropathies. Research was focused on autosomal recessive forms – demyelinating type CMT4C and axonal type CMT2B1. Most of the results obtained are related to these disorders. Data, which were obtained, are unique and were published in international journals with impact factor. Results obtained from CMT4C study are accepted for publication in *Clinical Genetics*. Results obtained in LMNA study (CMT2B1) were published in *Journal of Human Genetics*. The author performed and validated these new methods and original results, which are due to be used in genetic molecular testing of patients with hereditary neuropathies and muscle disorders:

1. Sequencing of all coding exons of the SH3TC2 gene. First mutations in the SH3TC2 gene in Czech HMSN I patients were found.
2. The prevalent mutation among Czech CMT4C patients was proven to be p.Arg954Stop.
3. Real-time PCR assay targeted at detection of the prevalent mutation p.Arg954Stop in the SH3TC2 gene was validated and is now used in our lab on a daily basis as a quick and efficient screening.
4. Molecular genetic testing of the SH3TC2 gene was introduced into the routine diagnostic testing scheme for Czech HMSN I patients.
5. Phenotype of CMT4C patients was inspected and described in great detail.
6. The LMNA gene sequencing in a group of Czech HMSN II patients.
7. Sequencing of the LMNA gene in 8 patients with Autosomal Dominant Emery-Dreifuss muscular dystrophy. Causal mutations were found in 3 patients (from two families). These are novel nonsense mutations.
8. Multiplex ligation-dependent probe amplification (MLPA) method was used for testing of 48 selected patients with HMSN II in whom no mutations in the LMNA gene were found....