

Sarcotubular Myopathy Due to Novel TRIM32 Mutation in Association with Multiple Sclerosis**Orest Semeryak***Lviv Regional Clinical Hospital, Ukraine*

Limb-girdle muscular dystrophies (LGMD) are a group of rare progressive genetic disorders that are characterized by wasting (atrophy) and weakness of the voluntary muscles of the hip and shoulder areas (limb-girdle area). Multiple sclerosis (MS) is a chronic neuroimmunologic (both the nervous system and the immunological system are involved) disorder of the central nervous system involving the brain, spinal cord and optic nerves. LGMD classification: The term limb-girdle muscular dystrophies is a general term that encompasses several disorders. These disorders can now be distinguished by genetic and protein analysis.. Autosomal dominant LGMD is known as LGMD1 and there are currently recognized eight subtypes (LGMD1A-1H). Autosomal recessive LGMD is known as LGMD2 and has 17 subtypes (LGMDA-Q).. Additionally, the problem of comorbidity is not covered in the literature, especially for multiple sclerosis (MS) and LGMDR8. Trim32: Tripartite motif-containing protein 32 (TRIM32) is a protein that ubiquitinates different substrates in muscle including sarcomeric proteins. Mutations in TRIM32 have been associated to Limb-Girdle Muscular Dystrophy 2H (LGMD2H) and Sarcotubular Myopathy, , now considered in the spectrum of LGMD2H. The Case: We describe a novel mutation in TRIM32 gene in an adult patient who was presented with the combination of MS and a moderate limb muscles weakness which was regarded as LGMDR8.

Biography

Orest Semeryak is a neurologist and pediatric neurologist, Chief of NGO «The Ukrainian Neuromuscular Diseases and Peripheral Nervous System Diseases Association», Director of Center of Neuromuscular Disorders in Lviv Regional Clinical Hospital. Having had graduated from Bukovinian State Medical Academy Ukraine and further studied the neuromuscular diseases in Mossakowski Medical Research Centre Polish Academy of Sciences, mentored by Irena Hausmanowa-Petrusewicz, followed by courses in the Institute of Myology of Paris he is now one of the most active popularizers of NM sciences in Ukraine.