ABSTRACT

Charles University in Prague
Faculty of Pharmacy in Hradec Králové
Department of Biological and Medical Sciences
Candidate: Bc. Monika Šrámková
Supervisor: Doc. MUDr. Josef Herink, DrSc.
Title of diploma thesis: Channelopathies of the nervous and musculoskeletal system

Channelopathies are rare group of mostly hereditary diseases that have one common trait – pathologically working ion channels. Ion channels are pores in plasmic membrane whose main function is ion transportation. They are also basis of nerve and muscle excitability. According to modern molecular methods there are known mutations of many genes of proteins of respective ion channels causing disruptions of their main functions via mechanisms of their loss, gain or change. Considering uncountable hypothetic mutations we can presume that other forms of this disease will be found. Channelopathies can affect any type of tissue but the most often they afflict nerve and muscle system. Clinical manifestation reflects impairment of functions of ion channels. In muscle tissue it leads to defect of contractile-relaxing cycle of muscles e.g. to paralyses or myotonias. In nerve tissue it leads e.g. to episodic attacks, myokymias, migraines and to other symptoms. Channelopathies are characterized by phenotypic and genotypic heterogeneity that may cause difficulty during diagnostics and invite to improvement in their knowledge. We can classify channelopathies according to affected tissue or in respect to type of defected ion channel. The most common are defects of potassium, calcium and sodium channels. To a group of muscle channelopaties belong e.g. hypercalemic periodic paralysis, congenital paramyotonia, malignant hyperthermia or congenital myastemic syndroms. To a group of nerve channelopathies belong episodic ataxia, familiar hemiplegic migraine or juvenile myoclonic epilepsy.