ABSTRACT

Desmin forms a muscle specific intermediate filament which participates in a formation of a dynamic intracellular network that links contractile apparatus with a sarcolemma, mitochondria and it provides a communication with a cell nucleus. This network serves to maintain morphological and functional aspects of muscle cells and cell organelles during a muscle contraction. Mutation in a desmin or an absence of desmin is causing a serious disease called desminopathy, which belongs among a group of myofibrillar myopathies. This disease manifests itself in all muscle types, however first observable defects occur in cardiac mitochondria, thus heart is a first organ manifesting symptoms of this disease. The purpose of this thesis was to summarize a current knowledge about physiological and molecular mechanisms that involved in a formation of intracellular desmin network and about patho-physiological states of desminopathies.

Key words: desmin, intermediate filaments, heart, desminopathy, cardiomyopathy