# Abstract

Huntington's disease (HD) is an autosomal dominant hereditary neurodegenerative disease characterized by motor, cognitive and behavioral disorders. HD is caused by expansion of CAG triplet (cytosine-adenosine-guanine) located in a gene on the short arm of the fourth chromosome. This expansion encodes an aberrant polyglutamine chain in the protein huntingtin. Physiological and mutated huntingtin (in case of HD) are expressed in almost all tissues and influences many cellular functions. The prevalence of HD in population is about 1 per 10.000. The disease is currently incurable and its mechanisms are not sufficiently understood. Besides affecting the central nervous system HD also affects peripheral tissues, including skeletal muscles. HD disrupts mitochondrial function and damages oxidative phosphorylation system, which has the task of producing energy in the form of ATP in cells. Research of transgenic minipig model for HD could help elucidate the mechanisms of disease‘s pathogenesis and potential therapeutic strategy.

In this diploma thesis, immunodetection with help of specific antibodies to detect changes in amount of 14 selected mitochondrial proteins in skeletal muscle tissue of three age groups of transgenic HD minipigs - 24, 36 and 48 months old was used. Gradual progression in reduced amounts of selected mitochondrial enzymes belonging to the oxidative phosphorlation and proteins associated with oxidative stress and mitochondrial structure destabilization was found increasing with age.



## Keywords:

Huntington‘s disease, huntingtin, mitochondria, oxidative phosphorylation system, OXPHOS, porcine model of Huntington's disease, transgenic model