Abstract

Huntington's disease is a neurodegenerative disease affecting the nervous system. It is caused by the mutation of the huntingtin protein coding gene. The mutation is manifested by the multiplication of CAG triplets.

Huntingtin is present more in the cytoplasm. It interacts with many proteins and has roles also in transcription and cell transport. Huntingtin also participates in correct regulation of embryonic development and development of nerve tissue. Mutant huntingtin causes oxidative stress, mitochondrial biogenesis and OXPHOS disorders.

Diagnosis of Huntington's disease is based on a laboratory examination of the presence of an allele predisposing to the disease. Clinical imaging is also an important part of diagnostics. The patient exhibits uncontrollable choreatic body movements and dementia.

The aim of the thesis was to describe the main characteristics of Huntington's disease with the focus of HD on mitochondrial energy metabolism. In the practical part, the aim was to analyze mitochondrial respiration by high resolution polarography in a set of 3 fibroblast lines from patients with Huntington's disease. Polarographic analyses showed that respiration of HD fibroblasts was decreased in comparison with controls. We found that complex II of oxidative phosphorylation was most affected in HD lines.

Key words: Huntington's disease, huntingtin, glutamine, OXPHOS