Abstract

The system of oxidative phosphorylation (OXPHOS) is main and essential source of energy (ATP) in eukaryotic cells. It is complex process situated in the inner mitochondrial membrane in which 4 enzymes of the respiratory chain, 2 mobile carriers and the ATP synthase participate. Defect in any part of OXPHOS may lead to the mitochondrial disorders.

An incidency of the mitochondrial disorders is estimated to be 1:5000 and mitochondrial disorders are the most common inherent metabolic diseases. Clinically, it is extremely heterogeneous group of disorders affecting primarily tissue with high energy demand, for example brain, hearth and muscle. With regard to broad spectrum of clinical symptoms and relatively poor genotype-phenotype correlation of mitochondrial genetic defects, it is preferred to perform complete laboratory diagnostic tests including several biochemical and molecular genetic approaches.

This bachelor thesis summarizes actual information about mechanism of OXPHOS and describes three major approaches to detect its functionality. Three major approaches are: spectrophotometric measurement of individual OXPHOS enzyme activities, measuring capacity of the mitochondrial energy generating system using various radioactive labeled substrates and measuring of oxygen consumption by polarography.

To achieve the best characterization of OXPHOS function, combination of these methods should be performed, preferably on two different tissues (usually skeletal muscle and cultured skin fibroblasts).