Abstract

Hereditary Mitochondrial Disorders Caused by Oxidative Phosphorylation Dysfunction

This bachelor thesis deals with problems of hereditary mitochondrial disorders that are caused by various dysfunctions of proteins in oxidative fosforylation known as OXPHOS complex.

Following recherche provides the reader with information about the genom and the structure of mitochondria where the OXPHOS is realized and about its structure and progress. I describe basic facts about various mutations in nuclear and mitochondrial DNA that negatively affect function of OXPHOS complex and the biogenesis of mitochondria.

The focus of this work is to summarize the newest data of individual diseases – their clinical manifestation, etiopatogenesis, prevalance in population and its possible treatment and prevention.

Key words: hereditary, mtDNA, mitochondrial diseases, OXPHOS complex