Abstract

Mitochondrial F_1F_O ATP synthase is a crucial enzyme that synthesizes the majority of cellular ATP and is part of a process called oxidative phosphorylation. This process is closely connected with important metabolic pathways in the cell, hence defects of the ATP synthase usually lead to serious metabolic disorders manifesting at a very early age. Based on the type of the enzyme defect, the mitochondrial function is affected leading to various changes in the cellular metabolism. The aim of this bachelor thesis is to summarize our knowledge about consequences of mitochondrial F_1F_O ATP synthase disorders reported in patients. Emphasis is placed on how these defects impact the function of mitochondria (e.g. the formation of the membrane potential or production of reactive oxygen species) and on the whole cellular metabolism.

Key words:

mitochondria, mitochondrial F_1F_O ATP synthase, cellular metabolism, mitochondrial disorders