Mitochondrial diseases / Mutations in mitochondrial DNA

Mitochondria are among the semi-autonomous cellular organelles. Each mitochondria contain its own genome in the form of *'circular mtDNA'* in 2-10 copies.

In mitochondria, the *citrate cycle* takes place; *Oxidative Phosphorylation* ; *& beta; -oxidation of fatty acids* ; *part urea cycle* . Mitochondria play an important role in apoptosis. Mitochondrial proliferation is found in highly metabolically active tissues such as skeletal muscle, heart muscle, brain, endocrine glands - these organs are particularly dependent on mitochondrial function.

In mitochondria, we encounter non-Mendelian "maternal inheritance." All mitochondria of the zygote come from the egg and are therefore passed on in the maternal line. The symptoms of the disease typically worsen with age (progressive course).

"Heteroplasmy" refers to a mixture of mutant and normal mtDNA molecules in the mitochondria of cells and tissues.

'Homoplasmy' means the presence of only mutant mtDNA.

Many of the diseases caused by mtDNA mutations are mitochondrial myopathies. In the muscle we find mitochondria of abnormal size and shape, which condition the appearance of rough red fibers (*ragged red fibers*).

Mitochondrial DNA is about ten times more susceptible to damage than nuclear DNA

- Mitochondrial DNA, unlike nuclear DNA, does not have the ability to repair
- there are many mitochondria in the cell, so when a cell divides, more mitochondrial DNA molecules have to replicate, increasing the risk of error,
- mitochondrial DNA has no histones,
- Mitochondrial DNA is very close to the respiratory chain and therefore to the reactive oxygen species that form during reactions in the respiratory chain, so it is more often oxidatively damaged.

Diseases related to oxidative phosphorylation disorder

Mitochondrial Disease / Respiratory Enzyme Deficiency

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