

The lactate level assessment in various body fluids plays an important role in the diagnostics of mitochondrial disorders in children. However, the interpretation of lactate level is often difficult due to its unspecificity and variability even in particular mitochondrial disorders. Three specific aims have been stated in this PhD Thesis: 1. To analyse the role of lactate examination in the differential diagnosis between children with mitochondrial disorders and children with other diseases. 2. To study the lactate level differences in various mitochondrial syndromes. 3. To characterise the clinical and laboratory data of neonates with mitochondrial disorders and to suggest new diagnostic algorithms. Clinical and laboratory data from patients hospitalized in the Department of Pediatrics were collected. Laboratory methods were provided in the cooperation with the Mitochondrial laboratory of the Department of Pediatrics and Institute of Inherited Metabolic Disorders. The study with lactate levels in 107 patients documented that brief seizures lasting less than 2 minutes did not increase lactate concentration in the CSF. CSF-lactate was a reliable marker in differential diagnosis in the children with mitochondrial disorders against children with epilepsy. 2. The severity of particular phenotype is more important for the severity of the lactic acidosis than the particular syndrome. 3. The diagnostic algorithms for neonates with suspicion of mitochondrial disorder were proposed. It is without any doubt that lactate is an important biochemical marker of children with mitochondrial disorder. This PhD Thesis evaluated some aspects of the role of lactate in mitochondrial disorders in order to help better interpret its values.