EXPANDED NEWBORN SCREENING (VNT 30)

Urea synthesis disorders

- Argininemia
- Argininosuccinic aciduria
- Citrullinemia
- Carbamoyl phosphate synthetase deficiency
- Ornithine transcarbamylase deficiency

Urea cycle converts highly toxic metabolism by-product ammonia to urea for excretion. Main diagnostic signs of these disorders are hyperammonemia and disturbed amino acid metabolism. Ammonia is neurotoxic and may cause acute encephalopathy, cerebral edema. This leads to lethargy, coma, and death.

Treatment: restriction of protein intake, reduced protein diet, ammonia binding drugs.

Amino acid metabolism disorders

- Hyperphenylalaninemia
- Hyperglycinemia
- Hyperhomocysteinemia
- Maple syrup in urine disease
- Tyrosinemia

Deficiency of various enzymes causes amino acid metabolism disorders, which lead to accumulation of toxic metabolites. It mostly damages brain, liver and kidney. Acute, unexplained health deterioration, progressive neurological symptoms, psychomotor developmental delay, intellectual disability may occur.

Treatment: specific diet, restriction of protein intake, specific amino acid mixtures.

Fatty acid beta-oxidation disorders

- Short-chain acyl-CoA dehydrogenase deficiency
- Medium-chain acyl-CoA dehydrogenase deficiency
- Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
- Very-long chain acyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Carnitine transporter deficiency
- Carnitine palmitoyl transferase I deficiency
- Carnitine palmitoyl transferase II deficiency
- Carnitine-acylcarnitine translocase deficiency

Fatty acid beta-oxidation – one of the main energy producing pathways, especially important while fasting. Due to these disorders toxic metabolites accumulate, fatty acids infiltrate liver, heart, skeletal muscle. Metabolic crisis is provoked by fasting, vomiting, infections with fever. It can lead to coma or sudden infant death.

Treatment: avoid fasting, frequent feeding, infusion with glucose, occasionally carnitine.

Organic acidurias

- Glutaric aciduria type II
- Glutaric aciduria type I
- Hydroxymethyl glutaric aciduria
- Holocarboxylase deficiency
- Isovaleric acidaemia
- 3-methylcrotonylglycinuria
- 3-methylglutaconic aciduria
- Methylmalonic acidemia
- Methylmalonic aciduria and homocystinuria
- Mitochondrial acetoacetyl-CoA thiolase deficiency
- Propionic academia

If protein metabolism is disturbed, organic acids accumulate and affect acid-base balance, metabolism of glucose, ketones and ammonia. Intoxication, vomiting, hypotonia, feeding difficulties, growth retardation, hepatosplenomegaly, acute or chronic encephalopathy, coma, sudden infant death may occur.

Treatment: specific diet, carnitine.