DIFFERENTIAL DIAGNOSIS OF ORGANIC ACIDEMIA: CLINICAL AND NEUROIMAGING FINDINGS

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Clinical differential Diagnosis

The organic acidemias are important in the differential diagnosis of metabolic and neurologic derangement in the neonate and of new-onset neurologic signs in the older child.

A-Organic aciduria

Several disorders, not classified as primary disorders of organic acid metabolism, have a characteristic urinary organic acid profile that suggests the appropriate diagnosis.

- Mevalonicaciduria, a disorder of cholesterol biosynthesis, shows mevalonic acid in the urine.
- Glutaricacidemia type II, a disorder of fatty acid oxidation, has multiple organic acids in abnormal concentration in urine. These organic acids include ethylmalonic acid, glutaric acid, dicarboxylic acids, and glycine conjugates of medium chain dicarboxylic acids.
- The fatty acylCoA-glycine conjugates that signal incomplete fatty acid oxidation and serves as signals to the diagnosis of MCAD defeciency and other disorders of fatty acid oxidation and transport.
- Biotinidase deficiency, a disorder of biotin recycling, results in the urinary excretion of several unusual organic acids, including 3-hydroxy-isovaleric, 3-hydroxypropionic, 3-hydroxybutyric acids, and acetoacetate. Propionyl glycine may also be seen.
- Mitochondrial diseases with disordered oxidative phosphorylation often demonstrate the presence of abnormal organic acids in the urine.

B-Acidosis

Non-genetic conditions, such as shock, sepsis, DKA, liver and kidney failure, thiamine deficiency, RTA, some drug intoxication cause acidosis- genetic conditions are include: inherited metabolic disorders of lactate and pyruvate metabolism and oxidative phosphorylation, disorders of the Krebs cycle such as fumarase deficiency.

C-Hyperammonemia

Disorders of the urea cycle and the hyperammonemia-hypoglycemia syndrome.

Neuroimaging

- A variety of **MRI abnormalities** have been described in the organic academia, including distinctive basal ganglia lesions in glutaricacidemia type I (GA I), white matter changes in maple syrup urine disease (MSUD), and abnormalities of the globus pallidus in methylmalonic acidemia. Macrocephaly is common in GA I.
- **Some differential agnosis** of MRI findings in organic academia is consist of: HIE, mucopolysacaridosis, middle fossa arachnoid cyst, leighdisease, hexachlorophene toxicity in neonates, myelin splitting disorders.
- Some organic aciduria such as L-2-Hydroxyglutaricaciduria may suggest leukodystrophy in MRI.

Keywords: Diferrential diagnosis; organicacidemia; children.

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