APPROACH TO ORGANIC ACIDEMIA

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Organic acidemias, also known as organic acidurias, are a group of disorders characterized by increased excretion of organic acids in urine. They result primarily from deficiencies of specific enzymes in the breakdown pathways of amino acids or from enzyme deficiencies in beta oxidation of fatty acids or carbohydrate metabolism. Organic acids also are found in the urine of some patients with mitochondrial disease.

Most organic acidemias become clinically apparent during the newborn period or early infancy. After an initial period of well-being, affected children develop a life-threatening episode of metabolic acidosis characterized by an increased anion gap. This presenting episode may be mistaken for sepsis, and if unrecognized, is associated with significant mortality.

Children with an organic acidemia are susceptible to metabolic decompensation during episodes of increased catabolism, such as intercurrent illness, trauma, or surgery. Parents and clinicians must be well informed about the initial signs of decompensation and trained in applying an emergency regimen . Surgeons and anesthesiologists should be aware of potential complications and their prevention during anesthesia and surgery.

Diagnosis has been facilitated through the use of gas chromatograph-mass spectrometry (GC-MS) and tandem mass spectrometry .Prenatal diagnosis is available for most disorders by detection of diagnostic compounds in amniotic fluid; by analysis of enzyme activities in amniocytes or chorionic villi; by molecular analysis; or by a combination of the three . Diagnosis also may be made through newborn screening by tandem mass spectrometry .

Laboratory findings are an essential part of the diagnostic approach to organic acidemias. In most organic acidemias, metabolism of glucose, ketone bodies, and ammonia is deranged primarily or secondarily, in addition to derangement of the acid-base balance. Hypoglycemia, lactic and/or ketoacidosis, and hyperammonemia of varying severity accompany the overt or compensated acidosis. In most instances, a definite diagnosis will be achieved by gas chromatography/mass spectrometry (GC/MS) studies of the urine.However sometimes definite diagnosis by clinical and laboratory assessments is not conclusive, in this cae diagnostic approach must be supported by loading tests.

The majority of organic acidemias may be treated by limiting the source of intake or removing the toxic intermediary metabolite. In the case of disorders lacking an effective treatment, an early diagnosis could lead to proper genetic counseling of the parents and to the option of reliable prenatal diagnosis of future pregnancies.

Keywords: Organic academia; diagnosis; approach.

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