Clinical outcome of organic acidemias – A twenty-five follow-up study in a reference center from Brazil.

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Introduction and Methods

Organic acidurias (OAs) are an important class of inherited metabolic disorders arising due to defect in intermediary metabolic pathways of carbohydrate, amino acids and fatty acid oxidation. More than 65 specific organic acids affecting these pathways have been identified.¹,² Maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), propionic aciduria (PA) and methylmalonic aciduria (MMA) represent the most commonly encountered abnormal organic acidurias.¹,² Glutaric aciduria Type I, classified as a 'cerebral organic aciduria. Long-term therapy is generally dependent on restricting precursors of the toxic organic acid which builds up because of the enzyme deficiency. The major clinical features are developmental delay, seizures, lethargy, coma, hypotonia, vomiting, failure to thrive, hepatomegaly, respiratory distress, and cardiac dysfunction.¹,² This is a 25 year (1997-2021) single reference center, cross-sectional, observational study which aims to describe the clinical status in 24 Brazilian OAs patients from 23 families.

References:
**Results**

Twenty-four patients (13 males: 11 females) were included with 7 types of OAs. Diagnoses were made on characteristic findings in urinary organic acid analysis. The major clinical features were developmental delay/intellectual disability in 19 patients (79.1%: 5 MSUD, 6 MMA, 2 GAI, 1 GAIi, 2 PA, 2 IVA, 1 DGA). Seizures in 20 patients (83.3%; 5 MSUD; 4 MMA, 1 GAI, 1 GAIi, 1 DGA, 2 IVA, 3 PA). Coma, hypotonia and vomiting were observed in 8 patients (33.3%; 5 MSUD; 1 MMA, GAI, 1 GAIi, 1). Hyperammonemia was seen in 10 patients (41.6%; 5 MSUD; 3 MMA and 2 PA). Death occurred in 4 (16.6%; 2 MSUD from liver transplant complications and 2 PA from metabolic decompensation). For all MMA patients, except for one MMA-B12 responder, the median methylmalonic acid level in urine was > 10,000 mmol/L. The treatment included L-carnitine, cofactors, and Metronidazole.
Early diagnosis and treatment allow the best chance of preventing clinical complications in patients with OAs. **Brazilians’ physicians need to think in rare diseases.**

Thank you =)