acid cycle and dicarboxylic acid shuttle by these compounds thereby supporting the notion of impaired energy metabolism playing an important role in the pathophysiology of cerebral organic acidurias.

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Impact of the chilean food complementary program (PNAC), in children with propionic (PA) and methylmalonic acidemia (MMA) first year results

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Introduction: PA occurs due to a deficiency of Propionyl CoA carboxylase enzyme and MMA due to a deficit of Methylmalonyl CoA Mutase or their cofactors. The treatment is a protein-restricted diet, special formula without methionine, threonine, valine and isoleucine (MTVI), supplementation with L-Carnitine, biotin and B12.
Objective: To determine the impact of formulas without MTVI subsidized by the PNAC.
Methodology: We compared pre and post PNAC data regarding calorie intake, protein, MTVI, calcium, iron, zinc, anthropometry (weight and height), ammonium level and acylcarnitines. Statistical analysis was performed using SPSS.
Results: 11 patients were admitted with PA and 4 MMA, age range between 2 and 18 years. There was a statistically significant difference in the contribution of special formula (average of 1 ± 0.7gr/kg/día increased) and a significant correlation with size. Also significant differences regarding supplementation of L-carnitine, MTVI and minerals. The size increased by 5.6 ± 4.4 cm and weight increased by 3.4 ± 5.4 kg, significant differences when compared with pre-admission.
Conclusion: Admission to the PNAC has positively impacted children with PA and MMA, reflected in the size and weight gain. However long-term evaluations are required.

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Primary hyperoxaluria type 1: organic aciduria diagnosed in plasma

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Background: Primary hyperoxaluria Type 1 (PH1) is a rare autosomal recessive inborn error of glyoxylate metabolism, caused by a deficiency of the liver-specific peroxisomal enzyme alanine:glyoxylate aminotransferase. The disorder results in overproduction and excessive urinary excretion of oxalate, causing recurrent urolithiasis and nephrocalcinosis.
Patient and methods: The patient, a 38 year-old-woman, was referred to our lab with severe arthritis in the hips and knees, calcinosis and stage V chronic renal failure under hemodialysis.
No urine samples were available to perform organic acids analysis so we studied patient plasma. Samples were extracted with ethylacetate and analyzed by GC-MS.
Results: Plasmatic organic acids profile in two different samples revealed a marked concentration of oxalate (131 and 125 μmol/L; controls: 0-5) and glycolate (362 and 338 μmol/L; controls: 9-42). Glycerate concentration was normal (17 and 15 μmol/L; controls: 0-24).
Conclusions: The usual biochemical indicator of PH1 is a persistently and markedly elevated urine oxalate. In the absence of urine samples, this biochemical diagnosis can also be done in plasma samples.
PH1 is a treatable organic aciduria and an early and accurate diagnosis preserves renal function of the patients. So, it is important to screen for PH1 in patients with recurrent urolithiasis or unexplained renal insufficiency.

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Magnetic resonance spectroscopy before and after encephalopathic crises in glutaric aciduria type I patients homozygous for the glutaryl-coa dehydrogenase IVS-1+5G>T mutation

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Background: Magnetic resonance spectroscopy (MRS) has been described as a potential functional non-invasive neuroimaging technique to detect neurologic striatal injury in patients with glutaric aciduria type I (GA-1). Decreased N-acetyl-aspartate/creatinine ratio (NAA/Cr) has been seen as a marker for reduction in neuronal integrity and neuronal loss, seen in the majority of GA-1 patients with encephalopathic crises.
Objectives: To understand the MRS abnormalities associated with striatal and white matter injury in GA-1 patients with the Manitoba variant of GA-1
Method: 8 Manitoba patients with GA-1 underwent a total of 11 single-voxel hydrogen MRS.
Results: 2 asymptomatic patients without encephalopathic crisis had normal MRS. Only 1 out of 3 patients in our cohort demonstrated decreased NAA/Cr on MRS during encephalopathic crisis. 3 more patients had a history of significant encephalopathic crises 7-15 years prior to their MRS and had normal NAA/Cr.
Conclusion: Although decreased NAA/Cr can be seen in MRS of GA-1 patients during an encephalopathic crisis, it does not appear to be a consistent feature in our small cohort. Contrary to previous hypothesis, the absence of decreased NAA/Cr in chronic symptomatic patients suggests that decreased NAA/Cr is a marker for acute biochemical disturbance rather than differential neuronal loss in GA-1.

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Clinical and genetic investigation of 6 Iranian cases of glutaric aciduria type1

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Background: Glutaric Aciduria type 1 is an autosomal recessive disorder caused by mutations in glutaryl-Co A dehydrogenase gene(GCDH). Although newborn screening can reduce adverse outcomes, it is not done in Iran. We investigated clinical and genetic aspects of 6 Iranian GA-1 patients.
Methods: The diagnosis was made by urinary organic acid and/or acylcarnitine analysis .Genomic DNA was extracted from peripheral blood lymphocytes.
Results: Six Patients'(4 ♂,2♀) mean diagnosis age was 6.5±1.6 yrs. All 4 couples were first cousin. Presentations: Macrocephaly6/6, Developemental Delay /Regression & Dystonia 4/6, Normal Development 2/6. MRI: Hydrocephaly 6/6, Frontotemporal Atrophy4/6, Temporal fossa arachnoid cysts2/6 . Mean urine Glutaric acid ,3-OH Glutaric acid levels: 2051.5± 1026,29.5±19.4 mmol/molcr. respectively. DNA analysis: 2 siblings homozygote 181G>C, One homozygote 881G>A, One heterozygote 1204C>T & 707T>C, the other 2 sibling results are pending. Except one previously reported mutation (1204C>T by Bierly et al 1996) others were new in Iran. After therapy among 4 symptomatic patients the heterozygote one had repeated metabolic acidosis attacks and others remained unchanged.
Conclusion: Our study indicates necessity of newborn screening to lower diagnosis age of GA-1 in Iran, also suggests that different presentation and treatment response may be due to mutation diversity which should be noted in therapy and genetic consultation.