029 - Creatine Metabolism and Hyperammonemia in Argentinian Patients With Ornithine Transcarbamilase Deficiency

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Introduction: Creatine (Cr) biosynthesis requires 2 enzymes, arginine–glycine amidinotransferase and guanidinoacetate methyltransferase, and it can be taken up by cells using transporters. Recent studies demonstrated the impact of toxic ammonia ($NH4^+$) in ornithine transcarbamilase deficiency (OTCD), a urea cycle defect (UCD), on Cr metabolism. The Cr secondary deficiency has been found in mice experimental models and in brain cells' primary culture but are yet to be tested in humans. **Objective:** To evaluate relationships between $NH4^+$ and Cr synthesis by guanidine compounds analysis in patients with OTCD. **Methodology:** A total of 6 patients with OTCD, 3 hemizygotes and 3 symptomatic carriers. The studies were performed in samples during crisis conditions. Measurement of guanidinoacetate (GAA) and Cr in urine were performed by gas chromatography; creatinine (Crn) in urine and $NH4^+$ in plasma by spectrophotometric methods. **Results:** In hemizygote patients with OCTD, GAA range was 0 to 11 mmol/mol Crn (controls: 2-220), with Cr in normal range, 28 to 1070 mmol/mol Crn (controls: 6-1208); all patients had severe hyperammonemia, 451 to 2182 µmol/L (controls: 10-47). In symptomatic carriers, the GAA range was 0 to 39 mmol/mol Crn with Cr concentration at lower limits of normal range, 44 to 70 mmol/mol Crn; patients had mild and severe hyperammonemia, 140 to 1093 µmol/L. **Conclusion:** According to this report, Cr metabolism is shown to be altered and may participate in central nervous system dysfunction in patients with UCD. Therefore, Cr supplementation should be a neuroprotective tool of toxic effect on brain exposed to ammonium in patients with UCD.

030 - Current Picture of Peroxisomal Diseases in Colombia From a Diagnostic Reference Center

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Introduction: The peroxisomal disorders are inherited diseases, resulting from alterations in the peroxisome biogenesis. Its incidence is estimated at 1:50,000 newborns alive, being the most common the X-linked adrenoleukodystrophy (X-ALD) with an incidence of 1/17.000, followed by Zellweger Syndrome (ZWS) and rhizomelic chondrodysplasia punctata (RCDP). **Objective:** To present the experience of the Institute of Inborn Errors of Metabolism at the Hospital Universitario San Ignacio in collaboration with the Kennedy Krieger Institute in the diagnosis of peroxisomal disorders between 2010-2013. **Methods:** Retrospective analysis of peroxisomal diseases diagnosis in Colombia. **Results:** 167 samples were evaluated and there was an increase in the number of ordering of this test (increase of 10.5 % related to the number in 2010). During this period, 7.2 % of the samples were positive (9 X-ALD, one ZWS, one RCDP, one elevation of triene/tetraene acids and 2 heterozygous X-ALD). Among the positive samples for X-ALD we identified two asymptomatic patients. The age of the onset of symptomatic patients was between 7-9 years and the main clinical signs were seizures and demyelination. **Conclusions:** There is no effective treatment for symptomatic patients with X-ALD but the use of Lorenzo's oil could lower very long chain fatty acids levels. The timely diagnosis also contributes to an adequate genetic counseling to families suffering from these diseases.

031 - Delayed Diagnosis of Nephrophatic Cystinosis in Mexico

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Background: Cystinosis is an autosomal recessive lysosomal disorder that may represent a challenging diagnosis because of low prevalence and symptoms similar to other diseases. **Objective:** To estimate the time elapsed between age of onset of clinical manifestations and age of diagnosis in Mexican patients with nephropathic cystinosis. **Methods:** Longitudinal observations of 36 patients with documented nephrophatic cystinosis. **Results:** A total of 36 patients (14 girls and 23 boys) from 25 families were diagnosed with nephrophatic cystinosis. In all, 34 patients were diagnosed with infantile cystinosis, and in 31 patients symptoms began before 2 years of age (15 before 6 months). All patients had Fanconi syndrome and failure to thrive, 33 had corneal crystals, and 30 rickets. A total of 24 patients were diagnosed after 7 years (1 month to 16 years) since the first clinical manifestation occurred. Four patients were diagnosed after kidney transplant. **Conclusion:** Cystinosis is a difficult disease to suspect, symptom onset is on average at 6 months, and diagnosis is delayed until 16 years in some