unspecific liver biopsy diagnosed as autoimmune hepatitis, initially responsive to prednisone and azathioprine. After 6 months, clinical worsening was evident and bile acid synthesis defect was suspected. The urine bile acid profile was consistent with 3β-HSD deficiency. Cholic acid was initiated followed by normalization of liver function. **Conclusion:** Defects of bile acid synthesis must be included in the differential diagnosis of cholestatic liver disease beyond neonatal period, because it is a good prognosis treatable defect.

014 - Biotinidase Deficiency: Assessment of Clinical and Molecular Aspects in a Sample of Brazilian Patients

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Introduction: Confirmation of biotinidase deficiency (BD), an autosomal recessive inborn error of metabolism, depends on enzyme activity measurement in plasma. Enzyme lability affects the identification of its true activity level (normal, heterozygous, partial deficiency, or total deficiency), making the decision about starting therapy difficult. **Objective:** To evaluate the clinical history and BTD gene sequence in Brazilian patients with BD. **Methods:** This is a cross-sectional multicenter study with convenience sampling. Clinical data and blood samples were obtained from 30 unrelated individuals with BD (15 male; 1 case of consanguineous parents) aged between 1 month and 18 years. Exons 2, 3, and 4 of BTD gene will be sequenced; exon 4 was the first. **Results:** The BD was identified by neonatal screening in 26 patients (23 are currently using biotin, and none shows symptoms) and based on clinical suspicion in 4 (most common manifestations: optic atrophy, motor regression, spastic paresis; onset of symptoms: 2 months-10 years of age; diagnosis: 7 months-18 years of age). Among the 19 patients whose exon 4 was analyzed, 6 different mutations and 1 synonymous substitution were found, all previously described in the literature. In 1 of 19 patients, no alteration was detected in the region analyzed. Mutation p.D444H (c.1330G>C) was the most frequent and was present in at least 1 allele in 15 of 19 patients. **Conclusion:** Our preliminary results suggest that there is a high prevalence of p.D444H in Brazil, which, according to the international literature, is the variant that is most frequently associated with partial BD.

015 - Nutritional Treatment for β-Ketothiolase Deficiency

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Background: β-ketothiolase deficiency is a mitochondrial defect in acetoacetyl-CoA thiolase in isoleucine catabolism. **Objective:** To present a patient with β-ketothiolase deficiency under nutritional management. **Case:** A 1-year-5-month-old boy who presented with jaundice at birth. At consultation he had diarrheal stools, vomiting, tachypnea, and anaphylactic breathing. When he started enteral feeding, new deterioration signs such as loss of sucking and swallowing, leading to gastrostomy were evident. In organic acids determination by GC/MS, 2 methylglutaconic, 3 hydroxyvaleric acid, 2-methyl 3-hydroxybutyrate, and triglilcarnitine were detected. Amino acid quantitation (high-performance liquid chromatography) showed high amino acids levels for alanine, methionine, serine, glycine, isoleucine, and valine, establishing the diagnosis of β-ketothiolase deficiency. Weight: 6.9 kg (3rd percentile), height: 79 cm (50th percentile), body mass index: 12.9 kg/m^2 (<3rd percentile), WBC: 12 cm. (<5th percentile) PCT: 6 mm. (5th percentile), C. Thigh: 19 cm, C. Cephalic: 47 cm. (25th percentile), P/T: −3SD, T/E: −1SD, P/E: −3SD. Nutritional management consisted of isoleucine-restricted diet and carnitine-formula supplement Ketonex providing 120 kcal/kg. **Results:** One year after treatment onset, the patient was 12 kg (50 percentile) in weight and height was 87 cm. Gastrostomy was removed, and amino acid levels were in the normal range 2 months later. **Conclusion:** Rapid diagnosis and monitoring of ongoing nutritional treatment is important for growth and proper development of patients with inborn errors of metabolism.

016 - Bone Disease as the Only Clinical Manifestation of Type I Gaucher Disease

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Introduction: Gaucher disease (GD) is a lysosomal disease, due to the deficiency of Î²-glucosidase, characterized by involvement of hematopoietic organs such as the spleen, liver, bone marrow, lung, and bone. Bone is the second most commonly affected structure, presenting bone infiltration and macrophage interleukins that determine osteoblast/osteoclast imbalance and a deleterious effect on bone. This impacts a patient's quality of life, causing pain, fracture, and orthopedic surgery requirements. We present a patient without hematologic or visceral involvement but with skeletal manifestations as the only clinical finding. **Objective:** To highlight bone disease as the main clinical manifestation in a patient with GD. To consider total body magnetic resonance imaging (MRI) scan as the most sensitive diagnostic tool in the assessment of skeletal involvement. **Patient:** C.K., a woman, 40 years old, was diagnosed with GD during a family study. Her sister was affected since she was 3 years, splenectomized at 18 years, and was under enzyme replacement therapy (ERT) for 14 years. After 10 years of follow-up without ERT, C.K. did not present visceromegaly, hematologic compromise, or crisis. Radiologic studies and bone density were assessed because of bone pain, showing normal results. Total body MRI showed severe skeletal involvement, with infiltration and osteolytic images in vertebrae, hip bones, and femurs. These findings associated with very high chitotriosidase lead to immediate ERT implementation. **Conclusion:** As bone is frequently affected in GD, it is important to perform skeletal studies using sensitive methods such as MRI to demonstrate affections and to determine the most appropriate therapy.

017 - Brain Macroangiopathy in Fabry Disease: Evidence by Magnetic Resonance Imaging

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Introduction: Fabry disease (FD) is a rare hereditary lysosomal storage disease that has been recognized as a possible etiology of stroke at a young age. Typical MRI findings are white matter lesions, microbleedings, vascular tortuosity (basilar dolichoectasia [BD]), Arnold-chiari type 1, and pulvinar sign. **Aim:** To present magnetic resonance imaging (MRI) findings in patients with FD from Argentina and relationship with the presence of cardiovascular risk factors (CVRFs). **Patients and methods:** A total of 70 patients with FD (27 men) were enrolled. We measured the presence of acroparesthesias, renal compromise, cardiac compromise, and corneal involvement. Enzyme replacement therapy (ERT) status was evaluated as well as the presence of cardiovascular risks factors. For BD, smoker's criteria were used. **Results:** Global mean age was 32.6 $\pm$ 1.8 years; 48% and 96.3% of women and men were under ERT, respectively. Renal compromise was reported in 60% of the population, cardiac compromise in 30%, corneal involvement in 91.4%, and acroparesthesias were present in 87.1%. Silent ischemias were found in 26% of the men and 30.2% of the women. The BD was the most frequent finding in men (63%) and women (39.5%). **Conclusion:** Our results show that cerebrovascular involvement may occur before the third decade of life. Presence of white matter lesions in MRI in young population is suggestive of FD. Given the high frequency of BD in young patients, this sign should increase FD suspicion by neuroradiologist.

018 - Broadening of Neonatal Screening in the Federal District of Brazil Through the Implementation of Tandem Mass Spectrometry

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Until 2008, the Brazilian Federal District neonatal screening program included phenylketonuria, congenital hypothyroidism, and hemoglobinopathies. In compliance with the Regional Law 4190/2008 that instituted a broadened neonatal screening protocol in the Federal District public network, tandem mass spectrometry was incorporated for screening of aminoacidopathies, beta oxidation defects, carnitine capture and transport defects, and organic acidemias. The objective of the current work is to describe mass spectrometry implementation in Federal District screening protocols. Cutoff establishment for diseases included in the broadened program for this population was carried out by a 3-step sampling process: first including 3500 samples (provided through collaboration with Dr Enzo Ranieri), then adding 9700 samples 5 months later, and finally 23 500 samples analyzed after another 7 months. Cutoff values were defined by calculation of 99th, 99.5th, and 99.9th percentiles for upper limits, and 0.1th and 0.5th percentiles for lower limits. Confirmatory tests for children with positive results were performed in partner laboratories using gold standard techniques. With the paradigm change in the data collection and analysis methods, after 2 years of experience and 90 000 samples analyzed, we have achieved 95.6% population coverage including children up to 7 years of age. Currently, we are the only Brazilian public service that performs neonatal screening for 22 diseases, together with the National Neonatal Screening Program.