Hb FCA, 02 Hb FC, 01 Hb FVA, 01 Hb FCD). Among those who were heterozygous, 14,224 Hb FAS, 2,259 Hb FAC, 456 Hb FAD and 435 carriers of hemoglobin rare variants were detected. For the study of rare variants, 42 DNA samples were obtained for sequencing and were characterized 23 alpha chain variants (3 Hb Woodville, 1 Hb Chad, 2 Hb Hasharon, 3 Hb G-Phil, 4 Hb G-Pest and 10 Hb Stanleyville) and 19 beta chain (11 Hb E-Sakatoon, 1 Hb Osu-Christiansborg, 1 Hb Richmond, 1 Hb O-Arab, 1 Hb J-Guantanamo, 1 Hb Shelby, 1 Hb Beckman, and 2 Hb Hope). **Conclusion:** Newborn screening allows early diagnosis of sickle cell syndromes and the inclusion of the carriers in prevention and treatment programs, reducing the morbidity and mortality in childhood. The variability of hemoglobin patterns identified in this sample reflects the heterogeneity of the southern population of Brazil. These data provide indicators that can be used in public health policies for improving the life quality of this population.

P080 - Anthropometric State Evaluation of Children Diagnosed With Maple Syrup Urine Disease (MSUD) Attended in a Reference Service in Newborn Screening in Salvador, Bahia, Brazil

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Introduction: MSUD is an inherited metabolic disorder associated to leucine, valine and isoleucine accumulation. Treatment consists in reducing serum concentrations of those amino acids, enabling affected children normal development and growth. **Objective:** Evaluate anthropometric status of MSUD patients treated with protein-restricted diet in Bahia. **Method:** Retrospective study with MSUD patients until July 2015. Data obtained from medical records review. Anthropometric status assessment used the indicators: height/age (H/A), weigh/age (W/A) and weight / height (W/A), to <05 years and for > 5 years the BMI / age (BMI/A) and height / age (H/A) with z-score value as the cutoff point according to WHO classification, 2006.Data was analyzed using EpiData software (v3.1). **Results:** Among nine patients studied, 55.6% (05) were girls with a mean age of 39 months (SD ± 34.78), ranging from 4 to 120 months. Six had MSUD classic form, two intermediate and one, non-determinate. Diagnostic age average was 23.38 days (SD ± 10.45) with symptoms onset mean age of 07 days (SD ± 4.04). One patient had severe short stature (H/A = -4.36) since admission and three had low height for age and all had adequacy W/H. At the end of the first year was observed worsening of the H/A with 71.42% (5/7) of severe short stature and 14.28% (1/7) of short stature, deficit W/H in two patients and one case of overweight. At the end of the second year there was improvement in growth, with only 50% (3/6) of short stature and adequacy of W/H for all the patients. At three years 01/04 child remained severe short stature and 02/04 short stature and all kept adequacy W/H. One child had obesity at 07 years of life keeping short stature. **Conclusion:** The disease has an important impact on linear growth already observed at the end of the first year of life. After this age, there is a growth recovery and maintenance of appropriateness of weight for height. This situation can be strongly associated with adequate nutritional therapy and multidisciplinary monitoring.

P081 - Clinical and Laboratory Characterization of Patients With Maple Syrup Urine Disease Followed in a Reference Service for Newborn Screening in Salvador, Bahia, Brazil

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Introduction: Maple Syrup Urine Disease (MSUD) is an inherited metabolic disorder of leucine, valine and isoleucine, whose accumulation leads to toxicity to central nervous system. Most frequent symptoms in the classic form are poor appetite, lethargy, neurological disorders, characteristic odor, seizures, hypothermia and coma. The Newborn Screening allows early diagnosis and treatment essentials to improve the clinical profiles. **Objective:** To characterize aspects of the diagnosis, treatment, and laboratory tests of MSUD patients attended at the Reference Service of Newborn Screening in Bahia-Brazil. **Method:** Retrospective study that evaluated patients diagnosed with MSUD until July 2015. Data obtained from medical records. Metabolic control tests were performed using the NeoLISA®MSUD kit and High Performance Liquid Chromatography (HPLC) and/or Tandem Mass Spectrometry (MS/MS). Data were analyzed using EpiData software (v3.1). **Results:** From nine patients studied, 55.6% (05) were female with average age of 39 months (SD ± 34.78), ranging from 4 to 120 months. Six had classical form of the disease, two intermediate form and in one the form was not determined. The average age of diagnosis was 23.38 days (± 10.45) with mean age of symptoms onset 07 days (± 4.04). Only 02 patients were diagnosed before developing clinical symptoms. The main symptoms at the first visit were poor suction (77.78%), seizures (66.67%), hypertonicity (55.56%) and skin lesions (55.56%). Seven patients were diagnosed by the combined dosage Leucine-Isoleucine with average of 1056.0 micromol/L (SD± 374.7) and median of 1083.0 micromol/L (755,0-1449.9), ranging from 427, 0 to 1479.8 micromol/L (VR 57.1 to 287 micromol/L). After completing six months of life was noted improvement in metabolic control in all patients.

Conclusion: Appropriate classification of clinical picture of disease and laboratory monitoring are important tools for clinical practice and dietary prescription for MSUD patients, allowing better matching of conducts to each patient tolerance.

P082 - Description of Nutritional Deficiencies in Children With Maple Syrup Disease (MSUD) in Dietary Treatment Attended in Reference Service of Newborn Screening in Salvador, Bahia, Brazil

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Introduction: MSUD is an inborn error of metabolism caused by deficiency of dehydrogenase enzyme complex activity that leads to leucine, valine and isoleucine tissue accumulation. Treatment consists of protein restriction and supplementation with metabolic formula valine, leucine and isoleucine free. It may lead to essentials vitamins and minerals deficient supply. **Objective:** To describe nutritional deficiencies or disorders in MSUD patients accompanied in reference service in Bahia-Brazil. **Methods:** Retrospective study analyzed MSUD patients until July 2015. Recommended criteria by WHO (WHO, 2011) was used to identify anemia, considering hemoglobin(Hb) <11.0g/dL in children up to 59 months of life, and Hb <11,5g/dL for children between 5 and 11 years. For vitamin and minerals dosage was considered laboratory cutoffs. Serum lipids assessment considered the reference values recommended by the I Directive of Atherosclerosis Prevention in Childhood and Adolescence (2005). Data were obtained from medical reports and analyzed using EpiData software (v3.1). **Results:** From nine patients, 55.6% (05) were female with an average age of 39 months (SD ± 34.78), ranging from 4-120 months. Six had classic form of MSUD, two intermediate and one non-determinate. Average diagnostic age was 23.38 days (SD ± 10.45) with average age of symptoms onset of 07 days (SD ± 4.04). Average hemoglobin was 11.6 (SD ± 1.2) g/dL ranging from 9,4-13,6g/dL, with three anemic patients, and one with hypochromic/microcytic anemia and normal ferritin. Four patients had creatinine low concentration with average of 0.38mg/dL (SD ± 0.05) and 50% (4/8) patients showed alkaline phosphatase elevation with average of 621U/L (SD ± 186.15) ranging from 300- 942U/L (VR to 645). None had hypoalbuminemia, folate, cyanocobalamin or vitamin D deficiency, abnormal levels of sodium or potassium. All evaluated patients had HDL low levels (VR>45mg/dLl). High values of triglycerides were found in 16.7%, and 50.0% had borderline values. **Conclusion:** Adequate dietary management prevents nutritional deficiencies. Although patients follow animal fat low diet and vegetable fat rich diet, it was noted changes in lipid profile, especially in relation to HDL levels.

P083 - Cystic Fibrosis Birth Prevalence Diagnosed by Expanded Neonatal Screening in Yucatan, Mexico

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Introduction: Yucatan is a peninsula located in south of Mexico and its population has a high degree of Mayan ancestry with low European admixture. An expanded newborn screening program that includes cystic fibrosis (CF) detection has been routinely performed in Yucatan since 2008. Although CF is a worldwide disease, traditionally it has been considered more prevalent in European descendants, but its prevalence in populations with indigenous Mayan ancestry is unknown. **Methods:** Immunoreactive trypsinogen (IRT) was determined through fluorometric immunoassay in dried blood spot samples collected from 100 public health centers distributed in the state of Yucatan, Mexico, from May 2008 to July 2015. All newborns with a suspicious result (cut-off IRT: >90 ng/mL) were retested. Sweat chloride tests were performed in those subjects with persistent IRT elevation (cut-off >60 mmol/L), along with a genetic mutation panel searching for the 32 most frequent CF mutations recommended by the ACMG. **Results:** The IRT determination performed on 71,888 newborns showed 132 suspected CF samples (0.18%); all suspected subjects were contacted and 131 of them accepted diagnostic protocol and follow up. Seven patients met the FQ diagnostic criteria and were confirmed after medical evaluation by an experienced pediatrician; 6/7 patients had *CFTR* molecular study, with p.[Phe508del] being the most frequent mutation (33.3%), followed by p.[Gly542Ter] (16.6%); in 50% of alleles, the pathogenic variation could not be identified in the recommended ACMG genetic mutation panel. **Conclusions:** The prevalence of CF in Yucatan, Mexico is 1:10,270 newborns, slightly higher than the reported for Hispanic Americans in the United States (1:13,500). CF is a worldwide disease that is also present in populations with Amerindian admixture. It is important for appropriate CF mutational analysis, to consider panels that includes the pathogenic *CFTR* variants described in the Mexican population by other authors, or using whole exome sequencing studies to identify all the mutations, including the new ones.

P084 - External Quality Assessment for the Detection of Phenylketonuria: Results of the Buenos Aires Programme

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