satisfactory and detectability by PCR and sequencing was successful in all the samples. In 10 from 12 samples, CAH diagnosis was confirmed while 2 patients turned out to be false positive cases, in agreement with molecular studies performed in DNA extracted from blood samples. **Conclusion:** Early confirmation of the diagnosis by molecular studies without the request of a second sample is very important to improve the clinical management of the patient, preventing salt-wasting crises and incorrect sex assignment. In this study we showed that DNA extracted from DBS is a useful tool for 21-hydroxylase deficiency diagnostic confirmation for newborn screening programs. Moreover, DBS represent an inexpensive method for long-term biobanking and for possible use in retrospective studies.

P014 - Evaluation Of Neolisa® MSUD Kit in Maple Syrup Urine Disease Child Monitoring in a Reference Service for Newborn Screening

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Introduction: MSUD patients require regular monitoring of serum leucine, isoleucine and valine, measured by high-performance liquid chromatography (HPLC), not easily available in Brazil public health services. NeoLISA® MSUD kit (MSUD kit) performance was analyzed, comparing it to HPLC. **Methods:** We evaluated medical records of 32 pairs of tests (MSUD kit/HPLC) obtained from 9 patients with MSUD treated in Bahia until July/2015. MSUD kit results expressed combined amounts of leucine and isoleucine (iso/leu) obtained from dried blood spot samples on filter paper. Leucine (leu) and isoleucine (iso) quantification was performed by HPLC in plasma collected in the same moment and forwarded to Inborn Errors of Metabolism Network Brasil (www.redeeimbrasil.ufrgs.br). They were classified as normal or elevated. Spearman correlation, kappa and predictive values were used in the comparison of the laboratories procedures. Iso/leu values (MSUD kit) were compared with leucine and isoleucine values obtained by HPLC, as well as the sum of leucine and isoleucine (iso + leu). **Results:** There were no differences in mean (SD) of iso + leu and leu [345.6 (175.1) versus 321.2 (517.0); p = 0802]. According to the kit MSUD, 12 (37.5%) of dosages were abnormal, while 13 (40.6%) of HPLC examinations showed leucine>216 micromol/L. Correlations observed between iso/leu values (MSUD kit) and leu, isoleu and iso + leu (HPLC) were 0.680; 0.530 and 0.746 (p <0.001), respectively. With the removal of a pair of outlier values (n=31), correlations were 0.649; 0.655 and 0.722, respectively. The kappa obtained was 0.241 (95%CI:-0.108-0.590), with 64.5% agreement. When considering abnormal values of leucine and/or isoleucine the kappa reached 0.323 (95% CI:0013-0633). In that case, the positive and negative predictive values were 75.0% and 60.0%, respectively. Among the tests with leucine above 500 micromol/L, MSUD kit identified 80% (4/5) of patients. **Conclusions:** The MSUD kit showed good correlation with the gold standard (HPLC) and good agreement, and may be useful for immediate assessment of MSUD patients, especially in more severe situations. However, HPLC testing remains mandatory.

P015 - Congenital Hypothyroidism. Progression of the Neonatal Screening Program in the Department of La Paz, Bolivia

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Introduction: Neonatal screening (NS) for congenital hypothyroidism was established by the laws of public health in Bolivia. Nevertheless, Bolivia does not have a national program. We present regional data of the Department of La Paz, with data from the Arco Iris Hospital, (private Hospital that applies public policies) and the Maternal-Infantile Hospital of the Social Security (CNS). **Objective:** To analyze the results of three years of congenital hypothyroidism (CH) screening and their impact in our health system. **Methods:** We included all newborns in the period of June 2012 to June 2015. Dried blood spot filter paper samples from a heel prick were analyzed for TSH levels, using the DELFIA (time-resolved immunofluorometric) assay. All TSH values above 10 μUI/mL were considered suspicious and were confirmed by a complete study of the thyroid (ultrasensible TSH, T3, T4 and free T4 by DELFIA). **Results:** 67000 newborns were screened in that period of time, 24 cases of congenital hypothyroidism were confirmed. The patients receive treatment and have periodical follow-up. The CH prevalence is 1:2792 with a coverage of 82% for the areas of service of the Arco Iris Hospital and Maternal-Infantile Hospital. **Conclusions:** Congenital hypothyroidism neonatal screening has been progressively increased with the incorporation of the Social Security, obtaining the mentioned coverage at the area of service of our two hospitals. Nevertheless, the official data of the number of births of the Department of La Paz shows approximately 53000 births per year in the province, and 73000 in the whole Department, for approximately 150000 births in the province and 22000 in the Department of La Paz. It is urgent to establish a comprehensive regional and national program for appropriate prevention of the intellectual disabilities associated with congenital hypothyroidism.

P016 - Distribution of the C.60+5G>T Danish Pathogenic Variant of PAH Gene Among Latin American PKU Patients and Phenotype Description of Resulting Homozygous State

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