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Introduction: The Neonatal Screening Program of the Province of Tucumán, Argentina, is headquartered in the Neonatal Screening Laboratory of the Institute of Maternity and Gynecology Ntra. Sra. de las Mercedes. It receives and processes dried blood spot filter paper samples for all the newborns (NB) in 13 hospitals of the 17 departments in the province. Congenital hypothyroidism (CH) is one of six congenital diseases screened for (CH, Congenital Adrenal Hyperplasia, Phenylketonuria, Cystic Fibrosis, Galactosemia and Biotinidase Deficiency). **Objectives:** To assess the prevalence of CH, and to evaluate comorbidities and sonographic features of thyroid glands in children; To assess a child's development through its height. **Patients and Methods:** A descriptive epidemiological study. Population Accessible included 139775 newborns whose samples were analyzed at the Neonatal Screening Laboratory in the period 2006-2014. The newborns with suspected CH were referred to Hospital of Niño Jesús for confirmation, treatment and monitoring. The clinical monitoring was conducted on a monthly during the first semester, every two months during of the second semester, and then every 3 months until to 6 years of life. From 7 years of life they received semiannual controls. Laboratory control was performed every two months during the first year of life. The growth control was made considering the SAP curves (Lejarraga et al). **Results:** Prevalence = $76/139775 = 5/10000$. Concomitant diseases: Down Syndrome 5/76, 20/76 with jaundice. Sonographic features of thyroid gland: 7% (increased), 18% (Hypoplastic), 24% (not displayed) and 7% (structural alteration). Height: 6 children with height value below the 3rd percentile. **Conclusion:** Early diagnosis and proper treatment allowed bone maturation and growth according to chronological age in the most of patients.

PI66 - 22 Years of Experience in Newborn Screening for Congenital Hypothyroidism

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Introduction: Newborn Screening (NBS) for Congenital Hypothyroidism (CH) was implemented by Fundación Bioquímica Argentina (FBA) together with Children's Hospital "Sor

María Ludovica" (HSML), in July/1992. Initially, NBS was carried out by request and without a structured program until the "Diagnostic and Treatment of Congenital Diseases Program" (Prodytec) was implemented by the Ministry of Health of Buenos Aires Province (BAP), on April/1995. **Objective:** To present the results of 22 years of experience working in NBS for CH. **Materials and Methods:** The functional organization of Prodytec includes: screening testing at FBA NBS Laboratory, and confirmation, diagnostic, treatment and follow-up at the HSML. Coverage is provided free of charge to all newborns (NB) born in public hospitals of BAP since July/2010. The screened NB population mainly belonged to BAP and in a lesser percentage to other provinces, but only those on the first group were attended and treated in the HSML. Sample collection was recommended between 24 h to 5th day of life, and TSH was measured using AutoDELFLIA Neonatal hTSH method (cut-off: 11.0 uU/mL). **Results and Discussion:** Until December/2013, 3,404,852 NB were screened, 85.6% belonging to BAP and 14.4% to other provinces, with a recall-rate of 0.14%. Diagnostic was confirmed in 1,567 children, thus determining a global incidence of 1:2,173 live births and showing a higher incidence in BAP regarding other provinces group (1:2,103 vs 1:2,708). Median (interquartile range) of age at sample collection and at screening result were 3 (2-6) and 12 (8-16) days for BAP group, and 5 (3-13) and 15 (10-24) days for other provinces group; while the age at diagnosis for BAP group was 18 (14-26) days. Reevaluation at 3 years of age was made at the HSML in 675 NB, being confirmed 644 (95.4%) as Permanent CH and 31 (4.6%) as transient forms. Etiologies of Permanent forms were: athyreosis 25.0%, ectopic disgenetic gland 57.1%, eutopic disgenetic gland 2.2% and eutopic thyroid gland 15.7%. About coverage, during 2013 Prodytec screened around 130.000 NB from public hospitals of BAP reaching a rate in this group > 97%. Until 2013, no one false negative result was reported.

PI67 - Relationship Between Anthropometric Development and Metabolic Control of Children With Diagnosis of Maple Syrup Urine Disease (MSUD) Monitored in a Reference Service in Newborn Screening (RSNS)

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Introduction: MSUD is caused by deficient activity of the enzymes responsible for metabolism of amino acids leucine, isoleucine and valine. Neonatal screening allows diagnosis and early treatment of patients. Treatment aims to reduce serum concentrations of leucine, valine and isoleucine in order to enable normal development and growth. **Objective:** To relate

the anthropometric development and metabolic control of children with MSUD followed at RSNS-Salvador/Bahia/Brazil. **Methods:** Retrospective study which evaluated patients diagnosed until July 2015. Data obtained from medical reports. Anthropometric status assessment used the indicators height/age (H/A), weight/age (W/A) and weight/height (W/H), for those under 05 years old, using value of z-score as a point cutting according to WHO classification, 2006. Metabolic control tests were performed using the NeoLISA[®]MSUD kit and high-performance liquid chromatography (HPLC). Data were analyzed using EpiData software (v3.1). **Results:** Among nine patients, five (55.6%) were female. They had mean(SD) age of 39(34.8) months, ranging from 4 to 120. Six had classical form of disease, two had intermediate form and one was not determined. Mean(SD) age of diagnosis was 23.4(19.4) days, with average(SD) age of onset of symptoms of 7.0(4.1) days. There was inverse correlation weak to moderate between the laboratory values serum of leucine, isoleucine and valine and W/H in the first three years of life, respectively, -0.275; -0.448 and -0.228. **Conclusions:** There was not a definite trend of association between laboratory values and anthropometric indicators W/H and H/A in the first five years, although an inverse relationship between these variables. This suggests a worse anthropometric development in presence of metabolic decompensation.

PI168 - Outcome in Patients With Phenylketonuria (PKU): What Should be Improved?

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Objective: To link adherence, neurological development and nutritional status of patients with phenylketonuria treated at a public hospital, identifying factors associated with poor outcome. **Materials and methods:** All patients with PKU treated at the Nutrition Department were included. They were classified, according to the value of phenylalanine achieved with usual diet, in severe, moderate, light PKU and persistent benign hyperphenylalaninemia. A patient was considered adherent to treatment if during the 1st year of life had 1 monthly control. 4 controls were considered appropriated in 1 to 10 patents and over 10, 3 controls were appropriated. We also took into account the average value of plasmatic phenylalanine to consider adherent to treatment: less than 360 for those under 13 and less than 600 for older children. The neurological development was evaluated considering school performance or neurologic evaluation for assistant physician. Nutritional status was determined by anthropometric control. Town of origin of the patient, and distance to specialized care center was recorded. **Results:** 20 patients were included, 5 with severe, 3 patients with moderate, 9 with light PKU and 3 persistent benign phenylketonuria. Considering all age groups 70% were adherents. 16 children had proper neurological development (80%) . 4

were considered neurologically compromised, 3 of them had severe PKU and were not adherent to treatment. However 2 of the severe PKU, adherent to treatment patients, had neurological development according to age. All patients had adequate nutritional status. All patients with poor adherence lived near de reference hospital and Just one patient live with only one parent. **Conclusion:** The nutritional status was adequate in all patients. Malnutrition didn't cause worse outcome. The distance between home and control center and living with only one parent didn't affected treatment adherence or outcome. Neurological development is compromised to a greater extent in patients with poor adherence, in accordance with the literature, and influenced by the type of PKU. Severe PKU have a higher incidence of neurological impairment. However 40% of patients with severe PKU have normal neurological exams. So the efforts of the medical and support team should focus on the monitoring of these patients especially, working with families, reevaluating strategies to improve adherence.

PI169 - The Role of the Psychologist as a facilitator of Mourning From Preparation Helping Improve Adherence to Treatment of Phenylketonuria

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Introduction: During life, and more intensely during pregnancy, expectant parents build an idealized image of a perfect son. The relationship between parents and children starts from that ideal. It is the relationship with the imaginary child who prepares the psyche of the country to establish ties with the real child who is to be born. The Neonatal Screening Program of Minas Gerais -PTN-MG, collects samples of blood on filter paper taken from the heel of newborns to diagnose genetic and metabolic diseases, including the Phenylketonuria (PKU). Grief has the function of preparing and psychic assimilation of the loss. In the case of a diagnosis of PKU, the child conceived no longer exists. Now there is a child with a chronic illness, full of limitations and differences, not the idealized love object. **Objectives:** To present the performance of the psychology team in supporting parents in the preparation of mourning the loss of the idealized son and his importance in the acceptance and compliance. **Methodology:** Through the methods of care, guidance and counseling conducted by Nupad - a reference center in neonatal screening Gerais- Gerais seeks to ease the grief process. We were met 22 families in the last 12 months, with an average of 05 calls per family. The sessions were weekly and made the psychology team. **Results:** Parents had great difficulties to get diagnosed. In that first moment the real baby is denied. Guilt, fear, anger, anxiety, anguish are