

levels in plasma by high performance liquid chromatography greater than 7 mg/dL. Confirmed cases were identified as benign hyperphenylalaninemia (levels 2 to 7 mg/dL), mild PKU (> 7 to 20 mg/dL), moderate PKU (> 20 to 30 mg/dL) and severe PKU (> 30 mg/dL). We used the SPSS statistical package to obtain the epidemiological indexes. **Results and Discussion:** 4,348,721 screening tests were made, with average coverage of 94.2%. 201 probable cases were identified, with index of suspicion of 0.005; and 43 were confirmed (PPV: 20.48%), the cumulative incidence was 1:101,133 live births. The diagnostic opportunity before 30 days was 23.3% and the date at which the final diagnosis was established was 52 days. Confirmed cases have only been reported in Aguascalientes, Baja California Sur, Chihuahua, Mexico City, Guanajuato, Guerrero, Jalisco, Mexico State, Nayarit, Nuevo Leon, Tabasco, Veracruz and Zacatecas. HPA benign cases were PKU 40%. **Conclusions:** The incidence of phenylketonuria found in our institution in Mexico is lower than reported internationally; we observed higher prevalence of cases in Bajío states. It is necessary to maintain an active surveillance system to improve the opportunity of diagnosis and treatment of patients.

**P055 - Hyperactivity: A Nearly Feature of Homocystinuria, Case Report**

Porras, L.(1); Sierra-ramirez, A.(2); Silvestre, J.(3); Munoz, N.(4)

(1): Clínica Comfamiliar Risaralda, Pereira, Colombia  
 (2): Ciencias  
 (3): Universidad de las Americas  
 (4): Metabolic Therapies

Inherited metabolic disorders comprehend a several diseases more commonly thoughtful in the setting of an acute illness with clinical decompensation. However they could have more insidiously presentation, with behavioral and psychiatric manifestations delaying the identification of the subacute disorder. Homocystinuria is a chronic and disable illness, which classically has been taken into account their manifestations ophthalmologic, cardiologic and skeletal as marfanoid habit and in general its systemic commitment, neglecting behavioral and psychiatric manifestations, which could be the initial feature of this disorder. In this case report, we are going to describe two siblings diagnosed with homocystinuria, both patients with behavioral disturbances and academic concerns. Patient number one, she was referred to a medical examination because academic concerns, unnoticed that her parents were of inherited metabolic disorder, despite that her parents were consanguineous and she had severe myopia. Only two years later, when she debuted with *ectopia lentis* the diagnoses was made, it showing as that this patients could be consulting for academic concerns without this kind of etiologies are taken into account. The diagnoses in patient number two is

**roduction:** The Social Security Institute of Peru (Essalud) in a pilot Newborn Screening (NBS) Program for congenital phenylketonuria and congenital adrenal hyperplasia at the Hospital Rebagliati Martins of Lima in October 2002, nationwide since January 2008. In the same year, screening phenylketonuria and galactosemia began only at the Rebagliati Hospital and its affiliated medical centers, and expanded Lima medical centers in 2010. In September 2011 the NBS became nationwide and included four diseases: congenital hypothyroidism, galactosemia, phenylketonuria and congenital adrenal hyperplasia, with a coverage of 100% of the insured population. The samples are processed by the Mother-Child Laboratory at the Edgardo Rebagliati Martins Hospital. Our objective is to report the incidence of Phenylketonuria in the Peruvian Social Security during the period 2012-2014. **Material methods:** Phenylalanine levels in newborns were measured fluorescent method (UMTEST) in dried blood spots collected on filter paper. The cut point was phenylalanine >3mg/>180 umol/L. A second assay was necessary to confirm positive cases. **Results:** NBS was performed to a total of 281,128 neonates with a coverage of 100%. A number of 9 were identified as probable cases, and 8 were confirmed. Incidence of PKU in the period 2012-2014 in Social Security was 1 in 35,141 live births. The confirmed cases were from metropolitan Lima (3), other provinces of Lima (2), Cusco (1) and Tumbes (1). **Conclusion:** The incidence observed in the Peruvian Social Security Institute (Essalud) was 141 live births, which is less than that reported in the international literature. This is the first report of PKU incidence in

**4 - Phenylketonuria Newborn Screening Experience in Mexico Epidemiological Review**

díaz Guerrero, J.(1); Delgado González, E.(1); Ortega Torres, M.(1); Mendiola Ramirez, K.(1); Ferrer Ica, L.(1)

Instituto Mexicano del Seguro Social, Coordinación de Atención a la Salud en el Primer Nivel, México D.F., Mexico

**Conclusion:** The neonatal screening to detect phenylketonuria was started in our institution in 2005. Approximately 100 newborns throughout the country are screened. Our neonatal screening program has a surveillance (SIVE) that identifies areas of opportunity and it is applicable to all states. **Objective:** This study was done to determine the coverage, index of suspicion, positive predictive cumulative incidence, opportunity of diagnosis and prevalence of means for Phenylketonuria (PKU) patients during the period from 2005 to 2014. **Methodology:** A cross-sectional study. Cases were identified by fluorescence (UMTEST) in dried blood spots, collected on filter paper. Probable cases were defined with the levels greater than 3 mg/dL (240 umol/L) and confirmed cases with the