P-185 - PREVALENCE OF HYPERPHENYLALANINEMIAS DETECTED BY NEWBORN SCREENING IN A CENTRALIZED LABORATORY FROM MEXICO.

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INTRODUCTION: In developed countries, newborn screening for hyperphenylalaninemia (HPA) has been carried out for more than fifty years; however, in emerging countries the detection of this disease has been gradually implemented. In Mexico, newborn screening for this disease is recent and HPA types according with the severity and its birth prevalence are poorly known. OBJECTIVE: To study the frequency of the different types of hyperphenylalaninemia according to the biochemical severity detected in a centralized laboratory from Mexico. MATERIALS AND METHODS: Retrospective analysis of the results of 1,898,096 newborn samples screened for HPA in a centralized laboratory from Mexico. Phenylalanine (Phe) was quantified using dried blood on filter paper by an enzymatic fluorometric method (GSP Neonatal Phe kit, Perkin Elmer®); HPA cases were defined as all those second samples whose Phe blood levels were above 120 µmol/L and low tyrosine (Tyr) levels with a Phe/Tyr ratio >2, determined by MS/MS. HPA types were classified as follow: benign HPA 120-360 µmol/L, clinically significant HPA >360-600 µmol/L, moderate phenylketonuria (PKU) >600-1,000 µmol/L, classic PKU >1,000 µmol/L. The geographic birth place of all HPA cases were also documented. RESULTS: 1,898,096 newborns were screened; 57 cases of HPA were confirmed in 24 of 32 states of the country. Benign HPA was found in 25 cases (44%), clinically significant HPA in 7 cases (12%), moderate PKU in 9 cases (16%) and classic PKU in 16 cases (28%), these last 16 cases were found in the states of Jalisco, Zacatecas, Queretaro, Guanajuato, Oaxaca, Durango, Nuevo Leon, Veracruz, Colima and Baja California. CONCLUSIONS: HPA were found in 1:33,300 screened NB and the classic PKU cases were localized mainly in the center occident of Mexico.