

O-016 - NEWBORN SCREENING PROGRAM FOR HEMOGLOBINOPATHIES IN THE STATE OF YUCATAN, MEXICO.

Sauri-Vivas M¹, Rodríguez-Rivero M², Ortiz-Aguirre SG³, Sáenz-Fuentes LG³, Manzanero-Traconis LG³, Peralta-Lazo AM⁴, Arias-Vidal CE⁴, Herrera-Pérez LA⁴, Maldonado-Solís FA⁴, Maldonado-Solís MA⁴

(1) Director general de los servicios de salud de Yucatán; (2) Departamento de equidad de género y salud reproductiva secretaría de salud - servicios de salud de Yucatán (SSY); (3) Unidad de salud materna y perinatal de secretaría de salud - servicios de salud de Yucatán (SSY); (4) TamizMas Screening Center de Químicos Maldonado, Yucatán, México.

INTRODUCTION: Hemoglobinopathies are qualitative or quantitative alterations of globin derived from genetic mutations whose consequences can be a structural modification or reduction of the synthesis of a globin chain (thalassemias). Around 400 variants of hemoglobin are known around the world and any chain can be affected: alpha, beta, gamma or delta. In the state of Yucatan, Mexico since 2012 an expanded neonatal screening program including the identification of these genetic variants was implemented. **OBJECTIVE:** To describe the frequency of hemoglobin variants found in the state of Yucatan, Mexico from 2012 to 2018. **MATERIALS AND METHODS:** Descriptive and observational study of the newborn screening for hemoglobinopathies accomplished in the period from 2012 to 2018. High performance liquids chromatography and isoelectric focusing were used to analyze blood samples obtained by heel puncture and collected on filter paper. **RESULTS:** 117,732 newborns (NBs) were screened and 697 were detected with a hemoglobin variant (686 carriers and 11 cases). Variants found were: 564 HbS (81%), 103 G-Philadelphia (15%), 18 HbC (2.6%), 4 β -variants (0.5%), 4 α -variants (0.5%) and 3 HbD (0.4%). The eleven affected NB cases correspond to: sickle cell disease (8), β -thalassemia (2) and HbC (1). **CONCLUSIONS:** The frequency of abnormal variants found in the state of Yucatan is 0.6%, being sickle cell disease the most prevalent one (1: 11,773 NBs).