

O-017 - EXPANDED NEWBORN SCREENING IN CHILE (26 CONDITIONS). RESULTS OF A PILOT PROGRAM.

Valiente AG¹, Faundez A¹, Cabello JF¹, Lobo G², Berrios CG², Valdebenito S², Cornejo V¹

(1) Instituto de Nutrición y Tecnología de los Alimentos (INTA), Universidad de Chile, Santiago-Chile. (2) Hospital San Juan de Dios (HSJD), Santiago-Chile.

INTRODUCTION: Since 1992, Chile implemented a national program of Newborn Screening (NBS) for Phenylketonuria (PKU) and Congenital Hypothyroidism (CH), preventing intellectual disability in more than 2,500 people. In 2015, the Ministry of Health (MH) of Chile proposed to expand the number of pathologies and in September 2017 INTA started the Pilot Program (PP) to screen 26 pathologies. The first stage of the PP was initiated at Hospital San Juan de Dios (HSJD), which runs the regional laboratory of the national PKU-CH program. **OBJECTIVES:** To present the implementation and results of the PP of NBS for 26 pathologies. **MATERIALS AND METHODS:** INTA trained medical and laboratory team of HSJD in preanalytical, analytical and postanalytical processes of the expanded NBS and analyzed samples of dried blood spots (DBS) of all the newborn at HSJD maternity. The applied techniques were: Tandem Mass Spectrometry (MS/MS), Fluorometry (FL) and Immunofluorometry (IFL). For FL and IFL commercial kits were used and for MS/MS "in house" non-derivatized technique was used. **RESULTS:** Of total 4.554 DBS analyzed, 3.752 (82.4%) corresponded to newborn of term. Recall rate was 1,6% (95% for MS/MS reanalysis). Samples were taken at mean of $1,7 \pm 0, 6$ days of life. Results reports were available at mean of $8,4 \pm 2, 2$ days of life. Of the total samples analyzed, 2/4.554 were positive. A case of a medium chain acyl-CoA dehydrogenase deficiency (MCAD) (diagnose at 29 days of age) and the second case a Glutaric Aciduria type-1 (diagnose at: 23 days of age) could be established. **CONCLUSIONS:** the excellent results obtained with the training of the staff of the HSJD, allowed a low recall rate and early diagnosis of 2 positive cases, and allows us to assert that this applied system can be reproduced all over the country. However, there are some challenges that must be considered within its expansion: the acquisition of adequate equipment capabilities, preparing human resources in these technologies and to modernize the registration system to implement the PP in a national scale.