P-222 - FIVE YEARS EXPERIENCE OF NEWBORN SCREENING FOR MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY.

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**INTRODUCTION:** The newborn screening for MCADD was added to our program after the set-up of the mass spectrometry (LC-MS/MS) technology in 2014. It is the seventh disease that is screened in our program. **OBJECTIVE:** to describe the results of the MCADD newborn screening at 5 years of its implementation. **MATERIALS AND METHODS:** Beginning in January 2014 MCADD screening was conducted in every newborn of our program. Blood samples were collected on filter paper Whatman 903 within 2-5 days of life. Non derivatized reagent from PerkinElmer and Chromsystems were used over the five years on API 3200 LC-MS/MS instrument from ABSciex. Primary marker used was Octanoylcarnitine (C8), cut off value of 0.28 uM (99.9 th percentile) Since July 2014 it was added the informative ratio C8/C10 with a cut off value of 1.0. Since that moment, babies with C8 and C8/C10 ratio above cut off values were recalled. Since January 2016, C8 cut off value of 0.21 uM and C8/C10 ratio of 1.0 using Chromsystems reagents were implemented. Confirmatory studies include acylcarnitines profile, Urine organic acids and molecular studies. **RESULTS:** The total number of newborns tested until December 2018 was 119,353, the total number of recalled babies was 31, Recall rate 0.026%. Five babies were confirmed with MCADD, positive predictive value at screening was 16.1 %. Mean days of life at newborn sampling was 2.8 days and mean days of life at physician visit for confirmatory testing was 9.2 days. All the patients were asymptomatic at their first physician visit and had good evolution. **CONCLUSION:** The introduction of this new disease at our program, allow the detection of patients with medium chain acyl coA dehydrogenase deficiency on a timely manner with acceptable recall rate and good evolution.