P-216 - PILOT STUDY FOR THE DETECTION OF CYSTIC FIBROSIS IN THE CUBAN NEONATAL SCREENING PROGRAM USING THE ULTRAMICROANALYTICAL SYSTEM

Castells-Martínez EM\(^1\), Sánchez-Gutiérrez A\(^1\), Frómeta-Suárez A\(^1\), Moksde Y\(^1\), Ozunas-Fernández N\(^1\), Licourt T\(^1\), Collazo T\(^2\), Rodríguez F\(^3\), Espinosa-Morales M\(^1\), Martín-González O\(^1\) et al.

(1) Immunoassay Center. (2) National Center of Medical Genetics. (3) Ministry of Public Health. Havana - Cuba. elisa.castells@cie.cu

INTRODUCTION: In Cuba no newborn screening program has been implemented for cystic fibrosis (CF). The UMELISA\textsuperscript{®} TIR Neonatal, has been developed for the measurement of immunoreactive trypsin (IRT) in dried blood spots on filter paper.

OBJECTIVE: To evaluate the analytical performance of the UMELISA\textsuperscript{®} TIR NEONATAL in the national network of SUMA laboratories. METHODS: Newborn dried blood spots (DBS) were evaluated in sixteen SUMA laboratories from several regions of the country. An IRT/IRT/DNA protocol was followed using a cut-off value of 50 ng/mL. DBS were analyzed to determine the influence of birth weight (BW), gestational age (GA), sex and sample processing time on IRT levels. A t-test was used to compare mean IRT concentrations among groups. A p-value <0.05 was considered as significant.

RESULTS: From January to June 2018, 6470 newborns were studied, obtaining a mean IRT value of 12.1 ng/mL (range 0-358 ng/mL) and a median of 9.0 ng/mL. Fifty-two samples (0.78%) were above the cut off level and sixteen samples (0.24%) were elevated again in the re-screening process. One of these samples was confirmed positive for CF by molecular biology technique (phe508del/c.3120+1G>A), constituting the first newborn screened and early diagnosed in Cuba. Samples were taken on average at 6 days and processed at 9 days. Second DBS samples were collected on average at 14 days and processed at 16 days of birth. Low BW (<2500 grams) and preterm babies (GA<37 weeks) had significantly lower IRT levels (n=518; 10.9 ng/mL and n=352; 10.4 ng/mL respectively). IRT concentrations in females (n=3023; 12.7 ng/mL) were statistically higher than in males (n=3290; 11.7 ng/mL). Lower IRT values were also observed in those samples processed after 10 days of collection (n=123; 10.2 ng/mL).

CONCLUSIONS: Sex, BW, GA and sample processing time lead to differences in IRT concentrations without consequences for the screening algorithm. However, because the sample size is small, further studies evaluating the influence of these factors must be carried out. The performance of UMELISA\textsuperscript{®} TIR Neonatal in the network of SUMA laboratories has been satisfactory, hence CF newborn screening was extended throughout the country from January 2019.