P-244 - EXPANDED NEWBORN SCREENING IN A POPULATION AT RISK OF VALLE DEL CAUCA- COLOMBIA

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INTRODUCTION: Inborn Errors of metabolism (IEM) are a set of conditions characterized by the accumulation of toxic substances usually produced by an enzymatic defect, that cause serious consequences in the newborn. Likewise, congenital hypothyroidism (CH), due to hormonal déficit, causes intellectual disability and dwarfism, a consequence that could be avoided with early detection. The incidence of IEM in Colombia is approximately 1/3000 live newborns and 50% develop the disease during the neonatal period. Their diagnosis and treatment has not been completely standardized, so the patients affected with these alterations are not being detected opportunely. The measurement of neonatal thyroid stimulating hormone (TSH) in umbilical cord for early detection of CH is the only legislated in the country but there is no infrastructure and equipment that allows to perform an expanded neonatal screening by tandem mass spectrometry, impeding the opportunity for newborns to have access to this program. OBJECTIVES: To check the usefulness and efficiency of expanded newborn screening using analytical tests for the detection of IEM. METHODS: Five Health Services Providing Institutions were included in the Department of Valle del Cauca-Colombia. 1000 baby samples were taken, with signs or symptoms that generated a medical suspicion of the presence of IEM. Blood samples were collected on filter paper for convenience, and analytical tests were performed to quantify TSH by immunoenzymatic tests, hyperphenylalaninemia by phenylalanine quantification by fluorometric ultramicroassay, Biotinidase deficiency by colorimetric ultramicroassay, congenital adrenal hyperplasia by quantification of 17-hydroxyprogesterone by immunoenzymatic tests and galactosemia test by fluorometric enzyme assay. RESULTS: The results of the tests carried out, showed that 2 newborn had TSH concentrations of 358 umol/L and 184.6 umol/L, respectively. In addition, 2 tests of phenylalanine with values of 497.2 umol/L and 289.6 umol/L were found. CONCLUSION: The increased in newborn screening by ultramicroanalytical tests allowed the early detection of patients affected by EIM. The patients in this study were confirmed and treatment was started. This technique is an effective and efficient alternative that, despite its limitations, achieves its objectives in the absence of most advanced technologies in our country.