P-223 - RAPID DETECTION OF A 3-HYDROXY-3-METHYLGLUTARYL-COA LYASE DEFICIENCY CASE THROUGH A SIMULTANEOUS NEONATAL SCREENING IN BLOOD AND URINE.

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INTRODUCTION: Traditional newborn screening for the detection of inborn errors of metabolism (IEM) is done analyzing dried blood samples on filter paper, however these require further analysis to confirm or discard the disease. An alternative to speed up the diagnosis could be the simultaneous analysis of dried blood and urine samples on filter paper. ***OBJECTIVE:*** To present a case of a 3-hydroxy-3-methylglutaryl-CoA lyase deficiency quickly diagnosed by simultaneous analysis of dried blood and urine on filter paper. ***MATERIALS AND METHODS:*** The dried blood sample impregnated on filter paper from a male newborn weighing 2,750 g, size 50 cm and 40 weeks of gestation was analyzed by Waters® MS-MS using a triple quadrupole and the dried urine sample on filter paper was analyzed by GC-MS of Agilent Technologies®. ***RESULTS:*** High concentrations of C5OH were found in the blood screening: 5.10 µmol/L (normal value < 0.64 µmol/L), C6DC: 0.21 µmol/L (normal value <0.24 µmol/L), C5OH/C8 ratio: 51.00 (normal value < 2.13), C5OH/C0 ratio: 0.43 (normal value < 0.01). In the urine, the following urinary organic acids were identified: 3-hydroxymethylglutaric acid, 3-methylglutaconic acid, 3-methylglutaric acid, 3-hydroxyisovaleric acid and 3-methylcrotonylglycine. Both results confirm the diagnosis of 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. The time from sample arrival to the laboratory until the notification of diagnosis was 3 days. ***CONCLUSIONS:*** IEM can be detected promptly with the simultaneous analysis of dried blood and urine samples on filter paper. The simultaneous analysis of samples in the neonatal screening could represent a great breakthrough alternative for the diagnosis of metabolic disorders in a shorter period of time and transcending the possibility of offering a better quality of life for screened newborns.