P-102 - IEM WITH GENETIC CO EXISTENT CONDITION. OUR EXPERIENCE WITH PKU AND OTC

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INTRODUCTION: Genetic co-existent conditions with IEM are rare, but have been reported specially with PKU. The identification and reporting of co-existent conditions are essential because it may alter morbidity and mortality of the IEM. Neuropsychological symptoms of an undiagnosed co-existent condition may be mistakenly attributed to the basic IEM. The association enhances the complexity of treatment and management burden for patients and caregivers of these cases.

AIM: to present the experience of our center with three cases of IEM with co-existent genetic condition: OTC and hyperphenyalaninemia with Down syndrome and PKU male with microdeletion of X chromosome. Cases 7 years old, male, PKU was diagnosed by NBS with good biochemical control with treatment started at 15 days of age. Behavior abnormalities were evident during the first year and BH4 deficiency was studied and discarded. An autism spectrum disorder and particular clinical dymorphism was evident at 2 years when CHG array was performed. A duplication of 1.7 bp in the Xp22.31 region was evident. This duplication is linked to pathological phenotype in a large number of cases associating intellectual disability. 8 years old, female, postnatal diagnosis of trisomy 21, with associated congenital heart disease surgically corrected at 7 months of age. At 4 years of age in the context of infectious respiratory disease, she presented encephalophatic episode with deterioration of consciousness, with plasma ammonium: 224 ug/dl, urinary organic acids showed elevated orotic acid and plasma aminoacids high glutamine. OTC gene sequencing showed known pathogenic variant. 6 years old, female, Down syndrome was evident at birth. Hyperphenylalaninemia was detected by NBS and remains stable during subsequent controls. CONCLUSIONS: when a case of PKU detected by NBS has a good biochemical control but the neurodevelopment is not normal, it is mandatory to look for other associated disease that explains the complex situation. Down syndrome is a frequent genetic disease and could be associated with rare IEM. An encephalopathic episode requires always measure ammonia no matter the known patient´s disease. When genetic condition co exists with treatable IEM, the patient and caregivers may need medical and social special support.