P-104 - THE USE OF MOLECULAR ANALYSIS FOR THE CONFIRMATION AND THE THERAPEUTIC ORIENTATION OF METABOLIC FINDINGS IN INBORN ERRORS OF METABOLISM.

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**INTRODUCTION:** The clinical manifestations of Inborn Errors of Metabolism (IEM) in the neonatal period can mimic other more frequent pathologies leading to possible confusions and delay in diagnosis, including the initiation of therapies that may hinder the specific metabolic investigation. **METHODS:** Describe the evolution of the patient in a Neonatal Intensive Care Unit, with a severe and complex condition, which despite altered metabolic examination, its diagnosis and therapeutics could only be safely confirmed after the molecular investigation of the newborn and your parents. **RESULTS:** Term newborn, first of the young non-consanguineous couple, mother with activity Lupus during the gestation in use of several immunosuppressors and pulmonary hypertension. On second day of life, the newborn presents a severe gastrointestinal bleeding with hypovolemic shock. He presented hepatomegaly, increased prothrombin time and aminotransferases. To control the clinical situation received transfusion of several blood products. On fourth day positive for Cytomegalovirus in the urine, started therapy with ganciclovir. After 2 days he maintained a clinical and had edema legs and ascites, in this moment a genetic evaluation was requested. On 8th day, the neonatal screening test (collected before blood transfusions), with an increase in galactose and a decrease in GALT activity, was received. Immediately introduced lactose-free diet and molecular analysis was performed on oral swab DNA sample, complete sequencing of the GALT gene was performed by Next Generation Sequencing (NGS). Two mutations were observed, pathogenic and probably pathogenic. Subsequently, the parents’ research revealed trans variants, identifying the molecular profile of the patient as a composite heterozygote for two variants in GALT. Corroborating the clinical diagnosis and the maintenance of the therapy. The evolution was only favorable with disappearance of edema with 30 days of life. **CONCLUSION:** In complex and rare diseases, the rapid recognition of the diagnostic possibility and parallel research of differentiation with common causes that can mimic it, are of the utmost importance. It is not uncommon for the affected patient to receive blood products, blocking out metabolic investigations. In this way, molecular research presents itself as a resolutive tool.