**INTRODUCTION:** The metabolome is the collection of small molecules (metabolites) found within a biological sample and metabolomics is the comprehensive study and analysis of these small molecules. In human plasma, the metabolites that circulate in the body represent products of complex metabolic pathways and their measurement can have clinical relevance in diseased patients. The human metabolome is influenced by genetics, environmental factors, epigenetics and the microbiome. Metabolomics technology can be applied to diagnosis of inborn metabolic diseases, and to follow up treatments.

**OBJECTIVE:** To describe the usefulness of metabolomic analysis in cases of patients with suspected neurometabolic disease, but with nonspecific manifestations.

**MATERIALS AND METHODS:** Untargeted metabolomics analysis were performed on patients evaluated in the genetics and neurogenetics services, in whom the clinical manifestations suggested the possibility of a metabolic disease, but the results of other biochemical tests, and neuro-radiology, did not allow to propose a specific diagnosis. The test, MetaETA IMDTM (Metabolon Inc., Morrisville, NC, USA), has demonstrated utility in the identification of biochemical disease signatures associated with a long list of inborn metabolic diseases. To further characterize this test, we performed correlation studies between the clinical and biochemical phenotypes.

**RESULTS:** We identified diseases clinically validated on the Meta IMDTM test: SCAD, MCAD, MGA, ABAT, THB, BH4, MEDNIK Syndrome, GA type 1, HMG-CoA (3-hydroxy-3-methylglutaryl-CoA) Lyase Deficiency, Trimethyllysine Hydroxylase Epsilon (TMLHE) Deficiency, and Citrullinemia. As result of complementary analysis, we confirmed a case of GMAT Deficiency, a condition that is currently clinically validated. We also are also in the process of confirming other pathologies that are not clinically validated. In addition, we are monitoring the treatment of a patient diagnosed with mutant PDP1 gene.

**CONCLUSION:** Metabolomic analysis can be employed as a powerful tool to study undiagnosed patients, mainly those with complex manifestations and non-conclusive findings in the traditional tests. Additionally, for patients with known diagnosis and under treatment, this test is useful for follow-up monitoring of efficacy.