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THE IMPORTANCE OF PLASMA AMINO ACID PROFILING IN THE DIAGNOSIS OF INBORN ERRORS OF METABOLISM

Hlistun Victoria, Efremov Egor, Blăniță Daniela, Boiciuc Chiril, Ușurelu Natalia
Institute of Mother and Child

Introduction:

Inborn errors of metabolism (IEM) make up a large group of disorders caused by an inherited defect of proteins that have enzymatic, carrier, receptor or structural roles. **Purpose:** The objective of the study was to appreciate the importance of plasma amino acids in the diagnosis of IEM

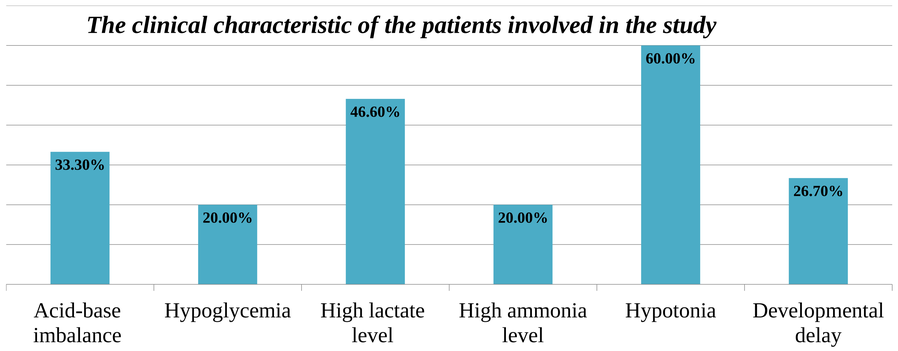
Keywords:

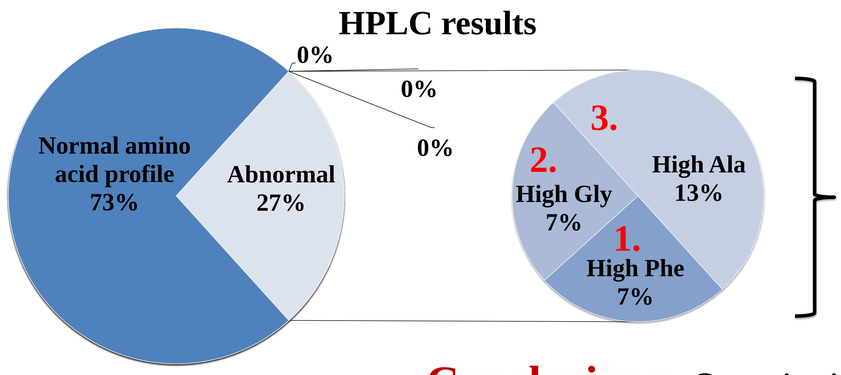
Amino acids, liquid chromatography, inborn errors of metabolism

Material and methods:

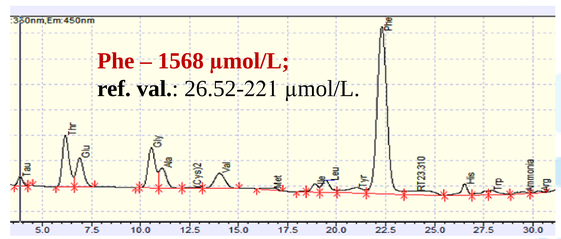
Plasma amino acids quantification was performed by high performance liquid chromatography (HLPC), Na-type, using Shimadzu LC-20 with post column derivatization with OPA and fluorescence detector, in 14 children with clinical signs specific for amino acid disorders.

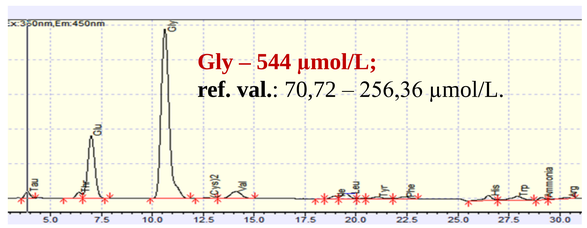
Results:





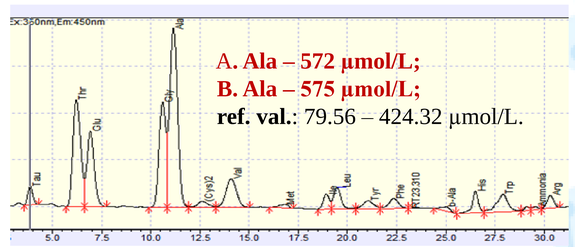
1. High phenilalanine (Phe) level have been identified in a 5 days old girl who had a familial history for Phenylketonuria (PKU). Her PKU screening results were abnormal being suggestive for disorders of Phe metabolism. The amino acid profile confirmed the suspicions.





2. High glycine (Gly) level have been seen in a girl of 2 months old, whose main clinical conditions were seizures from birth, precomatous state and respiratory arrest. Based on clinical features and metabolic results, Nonketotic hyperglycinemia (NKH) was suspected.

3. High level of alanine (Ala) has been observe in two patients. First patient (A) manifested frequent metabolic crises with severe lactic acidosis. The second patient (B) showed severe lactic acidosis, metabolic decompensation after virosis. The obtained data (high Ala level) suggest the mitochondrial involvement.



Conclusions: Quantitative analysis of amino acids is an essential step in the work-up for metabolic disorders. The early and precise diagnosis of IEM is essential for the long-term health of affected subjects.

No conflict of interests!