Journal of Nutrition and Metabolism

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Special Issue on

Amino Acids and Inherited Amino Acid-Related Disorders

CALL FOR PAPERS

Amino acids perform multiple essential physiological roles in humans, and, accordingly, their importance to health has been the subject of extensive attention. Inborn disorders affecting amino acid metabolism are generally rare but, by their very nature, complex and challenging conditions. Amino acid metabolism disorders comprise a very heterogeneous group of disease entities with a highly variable degree of onset and presentation. Clinical severity may range from incidental findings in some to overwhelming illness, brain damage, or multiorgan involvement in others.

Inborn amino acid-related disorders are generally caused by an inherited defect in the metabolic pathways, transport, or processing of a particular amino acid or a group of amino acids. Examples include Phenylketonuria (PKU), Maple Syrup Urine Disease (MSUD), Homocystinuria (HCU), and Urea Cycle Defects and Organic Acidurias, also called Organic Acidaemias, such as methylmalonic acidaemia (MMA).

Treatment goal for affected patients is to normalise the striking metabolic imbalance as much as possible by implementing dietary treatment and medication or cofactor supplementation along with patient monitoring and emergency treatment as required. In recent times, advances in diagnostic technology, including expanded newborn blood spot screening as well as major advances in treatments, have led to an exciting increase in the body of knowledge regarding amino acid-related disorders which will help to continuously improve our patient outcomes.

In this special issue, we will aim to provide some new insights into the patho-physiological roles of amino acids and treatment of inborn errors of amino acid metabolism in humans. We particularly welcome papers exploring novel therapies and patient outcomes, including interesting clinical studies, or focusing on the theoretical underpinning of such therapies.

Potential topics include but are not limited to the following:

- Rare inborn errors of amino acid/protein metabolism
- Diagnostic, patient management, and treatment
- Role of specific dietary amino acids in inborn disorders
- Novel approaches to therapy and clinical studies
- Newborn blood spot screening for inborn amino acid-related disorders
- Outcome of patients with inborn amino acid metabolism disorders
- Controversies and potential future directions

Authors can submit their manuscripts through the Manuscript Tracking System at http://mts.hindawi.com/submit/journals/jnme/aaia/.

Papers are published upon acceptance, regardless of the Special Issue publication date.

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