Journal of Pediatric Biochemistry 4 (2014) 181–182
DOI 10.3233/JPB-140124
IOS Press

Guest Editorial

New ideas in inborn errors of metabolism in children and infants

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In order to highlight the importance of inborn errors of metabolism (IEMs) in children and to update the journal readers regarding the state of the art in diagnosis, management and treatment, we have compiled a special issue of the *Journal of Pediatric Biochemistry* comprising various types of inborn errors of small molecules (organic acidemias), large molecules (lysosomal disorders), mtDNA disorders, and metabolic muscle disease. One additional section is devoted to educate the reader regarding how neuroimaging can be used to study the IEMs. Each expert shares current knowledge with the readers of this journal. This Special Issue will provide information on IEMs from a clinical, diagnostic, biochemical and therapeutic perspective.

The first article describes the use of neuroimaging in IEMs and stresses the importance of using a multimodal approach and illustrates the information that each type of imaging modality can provide to study and clinical diagnosis and monitoring of IEMs. For example, structural magnetic resonance imaging (MRI), which encompasses T1- and T2-weighted MRI, fluid attenuation inversion recovery (FLAIR), and voxel-based morphometry (VBM), is used to study brain's macroscopic structure, whereas functional magnetic resonance imaging (fMRI), which relies on oxygen extraction in areas metabolically active in response to a cognitive or motor task is used to study the neural nodes and networks underlying cognitive operations [1]. Diffusion weighted and diffusion tensor imaging (DWI and DTI) have utility in the study microstructural variance in white matter fiber tracts [2,3], and magnetic resonance spectroscopy (MRS) can visualize brain metabolism in static and dynamic models [4] and allow calculation of their concentrations.

Following this article, there are several contributions dealing with various categories of IEM. Kimberly Chapman, M.D., Ph.D. focuses on the identification, diagnosis, management and long term complications of the organic acidemias. This is accomplished not only by explaining symptoms, but also the medical decisions that need subsequently to be made in order to manage the patient and prevent neurologic and systemic injury due to intoxication with the metabolite in excess. This is followed by an article by Russell Saneto, D.O., Ph.D. who tackles the difficult diagnostic category of mitochondrial disorders due to mtDNA mutations and deletions. He defines the prototypical phenotypes and refers to diagnostic algorithms that are useful to the clinician.

Next in this issue, the reader will find a contribution by Pranoot Tanpaiboon, M.D. on the Lysosomal storage disorders. The author explains in detail the metabolic changes encountered in the various disorders demonstrating the similarities and differences and new advances in management and therapies.

This Special Issue concludes with one paper defining the diagnosis and distinctions between the metabolic myopathies and new therapies as well as those under development.

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In summary, this Special Issue on pediatric inborn errors of metabolism will be of value for the interested non-expert as well as for the knowledgeable biochemical geneticists and child neurologists. Compilation of a special edition addressing a group of rare diseases will hopefully raise awareness and promote early diagnosis with will improve the prognosis and health outcomes of affected patients.

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