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EDITORIAL

Disorders of the degradation of branched chain amino acids: What is new in clinics and laboratories?

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This issue of the Journal of Inherited Metabolic Diseases has special focus on genetic disorders of the catabolism of the branched chain amino acids leucine, isoleucine and valine. These conditions comprise classic organic acidurias such as propionic acidemia, isovaleric acidemia and methylmalonic acidemias as well as several less known inborn errors of metabolism. Most of the articles are based on key presentations of the 24th Annual Meeting of the German "Arbeitsgemeinschaft für pädiatrische Stoffwechselstörungen" (APS) held in Fulda in 2010. Abstracts of the 28 oral and 17 poster presentations at that meeting are available in Monatsschr Kinderheilkd 2010;158;293–304 [doi:10.1007/s00112-010-2181-9].

In the first article of this special issue Ronald Wanders and colleagues describe the biochemical basis of valine metabolism and disorders resulting from its impairment [doi:10.1007/s10545-010-9236-x]. Saskia Wortmann and coauthors provide a comprehensive overview including diagnostic guidance on the heterogeneous group of methylglutaconic acidurias which includes a disorder of leucine metabolism [doi:10.1007/s10545-010-9210-7]. Disorders of ketogenesis and ketolysis, which are introduced by Jörn Oliver Sass, comprise diseases that affect leucine and isoleucine metabolism [doi:10.1007/s10545-011-9324-6]. Ina Knerr and her colleagues address treatment options in the group of branched chain aminoacidopathies [doi:10.1007/s10545-010-9269-1]. In contrast, five more articles provide detailed information and new research findings on single disorders: Sarah Grünert and collaborators from metabolic centres in Germany, Austria, and Switzerland have compared the outcomes of propionic acidemia patients diagnosed following newborn screening and selective metabolic screening [doi:10.1007/s10545-011-9419-0]. Jan Kraus et al. have studied the mutations in the *PCCA* and *PCCB* genes of 54 members of that study population [doi:10.1007/s10545-011-9399-0]. Sabine Scholl-Bürgi and her colleagues focus on changes in amino acid metabolism in patients with propionic acidemia [doi:10.1007/s10545-010-9245-9].

William J. Zinnanti und Jelena Lazovic have used mouse models to investigate mechanisms of encephalopathy in maple syrup urine disease, a disease with impaired metabolism of the three branched chain amino acids valine, isoleucine and leucine [doi:10.1007/s10545-011-9333-5]. This collection of articles is completed by Johannes Zschocke's comprehensive overview on a disease initially recognized as a disorder of isoleucine metabolism due to methylhydroxybutyryl-coenzyme A dehydrogenase deficiency [doi:10.1007/s10545-011-9415-4]. As shown in his article, the issue is much more complex: The impairment of isoleucine metabolism appears not to be causal for the clinical course and *HSD10* disease may be a more appropriate nomination of this inborn error of metabolism.

In summary, this issue of the Journal of Inherited Metabolic Diseases presents updates on long-known metabolic diseases and in addition provides information on inborn errors of metabolism which have attracted little attention so far, although they are identified more and more frequently.

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