



JPR

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Editorial

Dear JPR readers,

We are so proud and happy to welcome you to the special inherited metabolic diseases issue of “*The Journal of Pediatric Research*”.

Inherited metabolic diseases are mostly inherited and occasionally *de novo* genetic disorders of the biosynthesis or breakdown of substances within specific pathways that were recognised by specific biochemical tests and sometimes treatable by metabolic intervention. In spite of known as “rare” disease, inborn error of metabolism, cumulatively affect approximately one in every 500 newborns. Therefore, they represent a special challenge in general and pediatric practice.

In the era of revolution in enzymatic and genetic diagnosis of inborn metabolic diseases this special issue is coming to illustrate and discuss the experience with enzymatic diagnosis in lysosomal storage disease, clinical findings and treatment in patients with Tyrosinemia Type I and different types of Mucopolysaccharidosis (Type II and IV). The readers will find the extensive knowledge for broad spectrum of inherited metabolic disease: from GM2- gangliosidosis, Niemann-Pick A and B, L-2-Hydroxyglutaric aciduria to Alkaptonuria. Interesting case reports will demonstrate the challenges in the diagnosis of patients with inherited metabolic disease: Glutaric aciduria Type I case with normal glutaryl carnitine, co-existence of two inherited metabolic disease in two patients and siblings with very rare disease: Ethylmalonic aciduria.

We would like to express deep gratitude to our authors, reviewers, editorial board members, Galenos Publishing House Officers and Nobel Pharmacy for their hard work in creating and support of this special issue.

Have a nice reading,

Best wishes

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