Special Issue

New Developments in Pediatric Clinical Genetics and Metabolic Diseases

Message from the Guest Editors

It is recognized that approximately 1 in 20 infants are born with a rare genetic condition, and of those infants, ~70% of these rare disorders begin in childhood (under the age of 18 years).

Around 20% of known conditions are due to disruptions of biochemical pathways leading to metabolic disorders that may have potential treatments. With the evolution of new techniques for the analysis of metabolites and the assessment of the function of biochemical pathways, in addition to the wider availability of whole exome and genome sequencing, our ability to recognize and diagnose disorders earlier than ever before and to detect milder presentations of conditions has grown immensely in recent years.

This Special Issue will provide an updated overview, novel insights, and critical perspectives on the pathophysiology, diagnosis, and treatment of rare inherited diseases in infants and children. Given the complexity and broadness of these topics, contributions from experts in the field through research papers and reviews are welcome.

Guest Editors

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Deadline for manuscript submissions

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Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the Genes team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider Genes for your next genetics paper?

Editor-in-Chief

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