Special Issue

Pediatric Rare Diseases: Genetics and Diagnosis

Message from the Guest Editor

Genetic factors are fundamental in diagnosing pediatric rare diseases, with about 70% presenting in childhood. Advances in genomic sequencing have improved diagnoses, but around 50% of pediatric cases remain undiagnosed. This highlights the need for more effective diagnostic tools, integrating genomics with other omics technologies. Referral to specialized genetics units and comprehensive phenotyping is essential for early interventions. The incorporation of artificial intelligence (AI), equitable access to genetic studies, and international collaborations are improving diagnostic precision. A correct diagnosis provides crucial information for families, enabling counseling, preventive strategies, and access to targeted therapies. In precision medicine, understanding genotype–phenotype correlations enhances individualized approaches, improving clinical outcomes for pediatric patients. This Special Issue, "Pediatric Rare Diseases: Genetics and Diagnosis," offers an updated perspective on genetic bases and diagnostic strategies for pediatric rare diseases, welcoming contributions on advancements, challenges, and opportunities in this evolving field.

Guest Editor

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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?

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