Special Issue

Current Diagnostics for Rare and Ultrarare Diseases

Message from the Guest Editor

The majority of an estimated eight thousand rare and ultrarare monogenic disorders have their onset in infancy and childhood. The repertoire of high-throughput genetic testing methods in routine and research is constantly expanding to include optical genomic mapping and short-read and long-read genome sequencing in all pediatric subspecialties. The finding of more than one genetic disease entity in the same patient or within a given family is an increasingly identified phenomenon, often adding to the observed intra- and interfamilial variability of the natural disease course and to the effect of therapeutic interventions. The interpretation of identified variants represents the main task in this field of human genetics. We welcome original research articles, case reports, and reviews addressing this topic, including the reporting of novel gene–disease associations and highly unusual variants and the segregation of one or more disorders in families.

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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