Special Issue

Next-Generation Sequencing in Rare Genetic Diseases

Message from the Guest Editors

Rare genetic diseases (RGDs) affect more than 300–400 million people worldwide. In the last decade, the advent of NGS (next-generation sequencing) and omics sciences, such as genomics, transcriptomics, and methylomics, has completely revolutionized the approach to RGDs.

This Special Issue aims to highlight the contribution of these novel approaches to unravel the pathogenetic mechanisms, discover novel disease genes and genotype–phenotype associations, and depict the genetic architecture underlying RGDs and driving novel therapeutic approaches. Original articles, case series, reviews, and descriptions of new methodologies in the field of RDs are welcome to contribute to this Special Issue.

Potential topics include, but are not limited to, the following: innovative approaches (NGS-based) to the diagnosis of RGDs (genomic medicine and multiomics data integration), big data and artificial intelligence, disease gene discovery, network analysis and rare disease (epi)signatures, including multilocus and mosaic disorders.

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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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