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Genes and Variants in Human Rare Genetic Diseases

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Message from the Guest Editor

Rare diseases (RDs) affect more than 300–400 million people worldwide, often causing chronic illness, severe disability, and premature death. In a large portion of cases, people with an undiagnosed disease incur in the so-called "diagnostic odyssey". An accurate diagnosis of an RD can result in better clinical management, the identification of potential therapeutics, and the avoidance of unnecessary treatments that may have non-negligible side effects. 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 RDs.

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

We look forward to receiving your contributions.

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Editor-in-Chief

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Message from the Editor-in-Chief

Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving 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.

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