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Genetics and Genomic Advances in Rare Diseases and Common Challenges

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Message from the Guest Editors

Genetic and genomic analyses have revolutionized clinical diagnosis for rare disorders, evolving from initial locus identifications to sophisticated technologies such as genome sequencing. Over the last decades, advances in the field have facilitated variant identification and interpretation across many rare diseases. However, these advances present new challenges.

This Special Issue aims to dissect the latest advances and hurdles in the genetics of rare diseases, emphasizing breakthroughs and limitations encountered in this field. Submissions are encouraged to focus on, but are not limited to, the following areas: advancements in genome sequencing techniques; novel genes and/or variants associated with rare diseases; insights from larger and more diverse disease/reference cohorts; challenges of non-coding variation; as well as the complexities stemming from limited or non-uniform phenotyping.

Authors are invited to submit reviews, original articles, and communications, aiming to pave the way for innovative solutions and advancements in the genetics of rare diseases.

Deadline for manuscript submissions:
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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.

Why not consider *Genes* for your next genetics paper?

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