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Molecular Genetics and Clinical Diagnosis of Rare Diseases

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Deadline for manuscript submissions:
closed (10 June 2024)

Message from the Guest Editor

Dear Colleagues,<false,>Advances in sequencing technologies have improved genetic diagnosis by enhancing the diagnostic yield, decreasing the time required to reach a diagnosis, and lowering the cost to patients and healthcare systems. Over the past decade, next-generation sequencing (NGS)-based disease-specific multi-gene panel, exome, and whole-genome sequencing have proven powerful in the diagnosis of patients with rare clinical conditions. In particular, the discovery of novel disease-associated genes has augmented our knowledge of the genetic etiologies of numerous previously undiagnosed human diseases, thus advancing our understanding of disease mechanisms and making it possible to develop potential treatments for these diseases.<false,>In this Special Issue, we welcome original articles and reviews covering aspects of new disease genes, novel methodologies for diagnosing rare genetic disorders, and the potential treatment of genetic diseases. These include, but are not limited to, studies related to novel disease gene discoveries, expanded disease phenotypes, the disease–gene relationship, novel disease mechanisms, treatments, and outcomes.

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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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