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Diagnosis of Rare Genetic Disorders

Guest Editor:

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Deadline for manuscript submissions:
closed (5 September 2023)

Message from the Guest Editor

Dear Colleagues,

Advances in sequencing technologies have improved genetic diagnosis. Over the past decade, next-generation sequencing (NGS)-based exome and genome sequencing have proven powerful in the diagnosis of patients with rare nonspecific clinical features. Particularly, discoveries of new disease genes have improved our knowledge of the genetic etiologies of many previously undiagnosed human diseases, which in turn advances our understanding of the disease mechanisms and makes it possible to develop potential treatment of the diseases.

In this Special Issue, we welcome original articles and reviews covering aspects of new disease genes, novel methodologies of diagnosing rare genetic disorders, and potential treatment of genetic diseases. These include, but are not limited to, studies related to novel disease gene discoveries, expanded disease phenotypes, disease–gene relationship, novel disease mechanisms, and treatment. We also welcome studies on methodologies relevant to the diagnosis of rare genetic disorders (lab approaches, functional studies, animal models, bioinformatics and artificial intelligence, causal analyses, and others).

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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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