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Molecular and Genetic Diagnosis of Rare Diseases

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Deadline for manuscript submissions:
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Message from the Guest Editors

An accurate diagnosis of rare diseases is essential in order to improve patient outcomes, provide access to specialized care and support, and increase quality of life (e.g., familial hypercholesterolemia or syndromic epilepsies). An understanding of the genetic and molecular basis of rare diseases helps to identify new therapeutic targets and develop novel treatments that not only patients with rare diseases can benefit from, but also those with more common conditions.

This Special Issue will first provide a comprehensive overview of the current state of knowledge and research in this field. Second, it will demonstrate the progress in technologies that are essential to studying the genetic background and pathomechanisms of rare diseases. Third, it will present examples of successful new diagnostic strategies for specific rare diseases and provide new clinical guidelines. Finally, this Special Issue will also be a platform for scholars to discuss emerging challenges and future prospects associated with the diagnosis of rare diseases.

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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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