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Genetics and Genomics of Rare Disorders Volume II

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Message from the Guest Editor

Since the first generation of DNA sequences, genetic analyses have gradually been introduced in clinical practice to support the diagnosis of rare disorders. Initially, loci and gene identification allowed geneticists to estimate transmission patterns and define recurrence risks of genetic Mendelian disorders.

To date, innovative molecular and cytogenetic technologies have been able to support diagnostic protocols, providing a timely diagnosis and early assistance. At the same time, the discovery of novel genetic etiologies for rare disorders is very important for the improvement of the diagnosis and genotype-phenotype definition, also supporting the development of novel therapies.

This Special Issue, entitled “Genetics and Genomics of Rare Disorders”, aims to present molecular and clinical aspects of rare genetic disorders. These include aspects related, but not limited, to the following topics: novel diagnostic approaches, genotype-phenotype correlations, application of research data into clinical practice, epigenetic approaches to rare disorders, functional studies, and animal models.

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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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