Special Issue

Molecular Genetics of Rare Disorders

Message from the Guest Editor

Rare diseases affect a small portion of the population, but collectively impact millions worldwide. Approximately 80% of rare diseases have a genetic basis, making molecular genetics a crucial field for understanding their etiologies, improving diagnoses, and developing targeted therapies. Advances in next-generation sequencing, genome-wide association studies, and functional genomics have transformed our approach to understanding these conditions and significantly enhanced our ability to identify disease-causing variants, uncover novel gene–disease associations, and elucidate pathogenic mechanisms. This Special Issue, Molecular Genetics of Rare Disorders, aims to highlight recent progress in the genetic and molecular characterization of rare disorders. We welcome original research and review articles covering novel disease gene discoveries, expanded phenotypic spectra, genotype–phenotype correlations, disease mechanisms, and therapeutic strategies. Contributions focusing on innovative diagnostic methodologies, including bioinformatics approaches, functional studies, and model systems, are also encouraged.

Guest Editor

Dr. Miriam Lucia Carriero
Department of Biomedicine and Prevention, University of Rome "Tor Vergata", 00133 Rome, Italy

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Genes
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
genes@mdpi.com

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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?

Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan
Department of Pathology, The University of Alabama at Birmingham, 1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, USA

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