Special Issue

Advance in Genomics of Rare Genetic Diseases

Message from the Guest Editors

Rare diseases are often characterized by disabling and life-threatening conditions, with few available treatment options. So far, about 10,000 rare diseases have been described and characterized but only few of them have been genetically solved. Therefore, the knowledge of the aetiology of a rare disease is crucial for the development and improvement of its treatment. Currently, recent and emerging technologies, such as high-throughput sequencing, provide unprecedented opportunity to clarify the aetiology of rare diseases, allowing genetic diagnosis and the development of new treatments. The aim of this issue is the collection of valuable contributions on the application of different genomic approaches, such as DNAseq, RNAseq or ChipSeq, for the characterization of the aetiology of rare diseases and the development of new therapeutic strategy, in order to ameliorate the clinical management of the affected patients. Moreover, the identification of new diagnostic biomarkers or the development of updated bioinformatics methodologies are topics of interest in this field.

Guest Editors

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Deadline for manuscript submissions

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Biomolecules is a multidisciplinary open-access journal that reports on all aspects of research related to biogenic substances, from small molecules to complex polymers. We invite manuscripts of high scientific quality that pertain to the diverse aspects relevant to organic molecules, irrespective of the biological question or methodology. We aim for a competent, fair peer review and rapid publication. Please look at some of the exciting work that has been published in Biomolecules so far. We would be delighted to welcome you as one of our authors.

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