# Special Issue

## Exploring Rare Diseases: Genetic, Genomic and Metabolomic Advances

### Message from the Guest Editors

Rare diseases, defined as life-threatening, chronically debilitating conditions, represent a substantial public health burden as they affect ca. 2–6% of the population. Currently, there are around 5000–8000 different rare diseases, and these numbers are continuing to increase. Despite intensive research, the genetic etiology and pathomechanisms of the majority of rare diseases are still unclear, and most of them do not yet have an approved therapy. Their diagnostics and care pathways are also challenging due to their rarity, heterogeneous manifestations, multisystem involvement and the often-observed incomplete penetrance. In addition, patients with undiagnosed genetic diseases often face a diagnostic odyssey that lasts for an average of eight years; moreover, a certain number of patients receive a misdiagnosis. The aim of this Special Issue is to collect original and review articles that provide cutting-edge knowledge related to genetic, genomic and metabolomic investigations in rare disorders. Keywords

- rare diseases
- genotype–phenotype analyses
- biomarker discovery
- molecular targeted therapy

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### Deadline for manuscript submissions

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The International Journal of Molecular Sciences (*IJMS*, ISSN 1422-0067) is an open access journal, which was established in 2000. The journal aims to provide a forum for scholarly research on a range of topics, including biochemistry, molecular and cell biology, molecular biophysics, molecular medicine, and all aspects of molecular research in chemistry. *IJMS* publishes both original research and review articles, and regularly publishes special issues to highlight advances at the cutting edge of research. We invite you to read recent articles published in *IJMS* and consider publishing your next paper with us.

## Editor-in-Chief

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