



Rare Monogenic Diseases: Molecular Pathophysiology and Novel Therapies

Guest Editor:

Dr. Ivano Condò

Department of Biomedicine and
Prevention, School of Medicine,
University of Rome Tor Vergata,
00133 Rome, Italy

Deadline for manuscript
submissions:

closed (15 September 2021)

Message from the Guest Editor

Most rare diseases we know arise from single gene mutations. In fact, the number of rare monogenic diseases is growing continuously, and to date, near 4000 single-gene inherited disorders have been characterized. Pathogenic mutations typically affect the coding regions, thus resulting in classical amino acid substitutions responsible for loss- or gain-of-function in protein products. However, several disease-causing defects originate from regulatory and non-coding DNA regions, ultimately affecting gene expression by transcriptional and/or post-transcriptional mechanisms.

Understanding the molecular pathophysiology of a rare monogenic disease has a double value. The identification of alterations that occur in specific genes, proteins, and pathways allows the translation of scientific advances into novel therapeutic approaches for these traits. Moreover, the investigation of rare monogenic diseases has the power to reveal fundamental biological mechanisms that would otherwise remain unknown.

This Special Issue will focus on the key molecular mechanisms that are affected within a rare monogenic disorder. New experimental therapies to target specific mechanisms are relevant.





an Open Access Journal by MDPI

Editor-in-Chief

Prof. Dr. Maurizio Battino

Department of
Odontostomatologic and
Specialized Clinical Sciences,
Sez-Biochimica, Faculty of
Medicine, Università Politecnica
delle Marche, Via Ranieri 65,
60100 Ancona, Italy

Message from the Editor-in-Chief

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.

Author Benefits

Open Access: free for readers, with article processing charges (APC) paid by authors or their institutions.

High Visibility: indexed within Scopus, SCIE (Web of Science), PubMed, PMC, MEDLINE, Embase, CAPlus / SciFinder, and other databases.

Journal Rank: JCR - Q1 (Biochemistry and Molecular Biology) / CiteScore - Q1 (Inorganic Chemistry)

Contact Us

*International Journal of Molecular
Sciences* Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

Tel: +41 61 683 77 34
www.mdpi.com

mdpi.com/journal/ijms
ijms@mdpi.com
X@IJMS_MDPI