



Rare Diseases - Molecular Mechanisms and Therapeutic Strategies (III)

Guest Editors:

Prof. Dr. Lidia Larizza

Experimental Research
Laboratory of Medical
Cytogenetics and Molecular
Genetics, IRCCS Istituto
Auxologico Italiano, Via Ariosto
13, 20145 Milan, Italy

**Prof. Dr. Maria Vittoria
Cubellis**

Dipartimento di Biologia,
Università Federico II, 80126
Napoli, Italy

Deadline for manuscript
submissions:

closed (31 December 2020)

Message from the Guest Editors

Dear Colleagues,

A rare disease is any disease that affects a small percentage of the population. The quantification of "small" is variable and, and represents an artificial border that will necessarily change with the diffusion of genetic screenings. More than 5000 rare diseases have been described. Non-sense mutations, deletions and insertions, abolish the function of the affected proteins, but mis-sense mutations have variable effects that go from complete inactivation to mild reduction of activity. At present, more than 70,000 mis-sense mutations have been reported. Taken together, these findings imply that there are different genotypes and phenotypes for any given disease. Bare figures give a flavor of the great challenge represented by rare diseases both in terms of diagnosis and therapy.

We are looking for papers that look into rare diseases with a genetic, biochemical or bioinformatic approach. Papers addressing specific pharmacological therapies for rare diseases are warmly welcomed.





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, CAPus / 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