

Special Issue

Rare Diseases: Molecular Mechanisms and Therapeutic Strategies

Message from the Guest Editors

A rare disease is any disease that affects a small percentage of the population. The quantification of "small" is variable and, in any case, 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.

Guest Editors

Prof. Dr. Maria Vittoria Cubellis

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

Prof. Dr. Lidia Larizza

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

Deadline for manuscript submissions

closed (30 April 2018)



International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 8.1
Indexed in PubMed



mdpi.com/si/8948

*International Journal of
Molecular Sciences*
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
ijms@mdpi.com

mdpi.com/journal/

[ijms](https://ijms.mdpi.com)





International Journal of Molecular Sciences

an Open Access Journal
by MDPI

Impact Factor 4.9
CiteScore 8.1
Indexed in PubMed



[mdpi.com/journal/](https://mdpi.com/journal/ijms)

ijms



About the Journal

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.

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

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)