

Special Issue

Genetics Research of Rare Human Diseases

Message from the Guest Editor

Inherited rare genetic disorders are, by definition, low-frequency diseases, but since thousands of different diseases exist, millions of people around the world are affected. Rare diseases can be a powerful tool in understanding new molecular pathways since they can present a high grade of heterogeneity (both in genetics and/or in phenotype). The aim of this Special Issue is to highlight new molecular and cellular mechanisms underlying these congenital diseases, as well as new therapeutic targets and/or treatments. We cordially invite authors in the field to submit reviews or original research articles highlighting new advances in understanding and treating rare diseases.

- inherited genetic diseases
- rare diseases
- molecular mechanisms
- cellular mechanisms
- therapy

Guest Editor

Dr. Lisa Ganesello

Clinical Medicine 1, Department of Medicine, University of Padua, Padua, Italy

Deadline for manuscript submissions

closed (31 January 2023)



Biomedicines

an Open Access Journal
by MDPI

Impact Factor 3.9
CiteScore 6.8
Indexed in PubMed



mdpi.com/si/114071

Biomedicines
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
biomedicines@mdpi.com

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





Biomedicines

an Open Access Journal
by MDPI

Impact Factor 3.9
CiteScore 6.8
Indexed in PubMed



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



About the Journal

Message from the Editor-in-Chief

Biomedicines (ISSN 2227-9059) is an open access journal devoted to all aspects of research on human health and disease, the discovery and characterization of new therapeutic targets, therapeutic strategies, and research of naturally driven biomedicines, pharmaceuticals, and biopharmaceutical products. Topics include pathogenesis mechanisms of diseases, translational medical research, biomaterial in biomedical research, natural bioactive molecules, biologics, vaccines, gene therapies, cell-based therapies, targeted specific antibodies, recombinant therapeutic proteins, nanobiotechnology driven products, targeted therapy, bioimaging, biosensors, biomarkers, and biosimilars. The journal is open for publication of studies conducted at the basic science and preclinical research levels. We invite you to consider submitting your work to *Biomedicines*, be it original research, review articles, or developing Special Issues of current key topics.

Editor-in-Chief

Prof. Dr. Felipe Fregni

1. Neuromodulation Center and Center for Clinical Research Learning, Spaulding Rehabilitation Hospital and Massachusetts General Hospital, Harvard Medical School, Boston, MA 02114, USA
2. Department of Epidemiology, Harvard T.H. Chan School of Public Health, Boston, MA 02115, USA

Author Benefits

High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, PMC, CAPLUS / SciFinder, and other databases.

Journal Rank:

JCR - Q1 (Pharmacology and Pharmacy) / CiteScore - Q1 (Medicine (miscellaneous))

Rapid Publication:

manuscripts are peer-reviewed and a first decision is provided to authors approximately 17 days after submission; acceptance to publication is undertaken in 2.9 days (median values for papers published in this journal in the first half of 2025).