



an Open Access Journal by MDPI

## Genetics Research of Rare Human Diseases

Guest Editor:

**Dr. Lisa Gianesello**

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

Deadline for manuscript  
submissions:

**closed (31 January 2023)**

### Message from the Guest Editor

Dear Colleagues,

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



[mdpi.com/si/114071](https://mdpi.com/si/114071)

# Special Issue



an Open Access Journal by MDPI

## 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

## 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.

## 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, CAPlus / SciFinder, and other databases.

**Journal Rank:** JCR - Q1 (Pharmacology and Pharmacy) / CiteScore - Q1 (Medicine (miscellaneous))

## Contact Us

*Biomedicines* Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland

Tel: +41 61 683 77 34  
[www.mdpi.com](http://www.mdpi.com)

[mdpi.com/journal/biomedicines](http://mdpi.com/journal/biomedicines)  
[biomedicines@mdpi.com](mailto:biomedicines@mdpi.com)  
[X@Biomed\\_MDPI](https://twitter.com/Biomed_MDPI)