# **Special Issue**

## Human Genome Sequencing in Health and Disease

## Message from the Guest Editors

This Special Issue on "Human Genome Sequencing in Health and Disease" will mainly focus on the following trends, addressed as reviews, new methodological approaches and original research:

- Whole genome sequencing developments in the identification of CNVs and trinucleotide repeat expansion
- Improving the interpretation of genomic data
- Genomic-based cancer diagnosis and treatment
- Diagnosing neurodevelopment disorders
- RNA vs. DNA sequencing: strategies to improve diagnostic yield
- Diagnosis and treatment of rare hereditary diseases
- Unraveling missing hereditability with genomic sequencing
- Neomutations, gonadal mosaicism or parent low-level mosaicism: new perspectives from deep sequencing
- Pharmacogenomics in clinical practice: are we ready?
- Liquid biopsy: current state and future perspectives
- Transcriptomic applications in cancer research and in clinical practice

#### **Guest Editors**

Prof. Dr. Fernando Regateiro

Institute of Medical Genetics/UC Genomics, Faculty of Medicine, University of Coimbra, 3000-548 Coimbra, Portugal

Dr. Henriqueta Silva

Institute of Medical Genetics/UC Genomics, Faculty of Medicine, University of Coimbra, 3000-548 Coimbra, Portugal

### Deadline for manuscript submissions

closed (31 January 2024)



an Open Access Journal by MDPI

Impact Factor 3.9
CiteScore 6.8
Indexed in PubMed



mdpi.com/si/180590

Biomedicines
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +4161 683 77 34
biomedicines@mdpi.com

mdpi.com/journal/biomedicines





an Open Access Journal by MDPI

Impact Factor 3.9 CiteScore 6.8 Indexed in PubMed





## **About the Journal**

## Message from the Editor-in-Chief

Biomedicines (ISSN 2227-9059) is an open access iournal 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

- 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).