biomedicines

IMPACT FACTOR 3.9 | Indexed in: PubMed | CITESCORE 5.2

an Open Access Journal by MDPI

Rare Genetic Diseases: From Pathophysiology to Novel Therapeutic Approaches

Guest Editor:

Dr. Doris Hendig
Institut für Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitätsklinik der Ruhr-Universität Bochum, Bad Oeynhausen, Germany

Deadline for manuscript submissions:
closed (30 September 2021)

Message from the Guest Editor

Dear Colleagues,

Studies on rare genetic diseases can help to get a better understanding of various molecular mechanisms and the development of novel therapeutic approaches even for disorders that are more common. This Special Issue will focus on the pathophysiology of rare genetic disorders, such as Pseudoxanthoma elasticum (PXE) or related disorders. Although much knowledge on the pathophysiology of PXE and ABCC6 has been gathered in the last two decades, we still do not know the primary pathomechanisms leading to its manifestation, as well as the physiological function of the encoded transporter protein ABCC6. Different therapeutic approaches have reached clinical studies, but none of them have been able to thoroughly prevent or cure PXE. This Special Issue aims to present an update on the basic insights into the pathophysiology along with novel aspects of the clinical presentation and diagnostics as well as therapeutic approaches of rare genetic diseases, such as PXE or others. We invite authors working in the field to submit original research articles as well as reviews fitting the scope of this Special Issue.

Dr. Doris Hendig
Guest Editor

biomedicines

IMPACT FACTOR 3.9 | Indexed in: PubMed | CITESCORE 5.2

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 - Q2 (*Medicine (miscellaneous)*)

Contact Us

Biomedicines Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

Tel: +41 61 683 77 34
www.mdpi.com

mdpi.com/journal/biomedicines
biomedicines@mdpi.com
𝕏@Biomed_MDPI