

Special Issue

Rare Genetic Diseases: From Pathophysiology to Novel Therapeutic Approaches

Message from the Guest Editor

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.

Guest Editor

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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.

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