children

an Open Access Journal by MDPI

IMPACT FACTOR 2.0

Indexed in: PubMed

CITESCORE 2.7

"Rare or Not So Rare, That Is the Question": Orphan Diseases in Children

Guest Editor:

Dr. Wilfredo De Jesús Rojas
Department of Pediatrics, University of Puerto Rico, San Juan, PR 00921, USA

Deadline for manuscript submissions:
closed (20 May 2022)

Message from the Guest Editor

More than 6000 rare diseases affect more than 25 million of individuals in the United States of America. A disease that affects less than 200,000 individuals is considered to be a rare disease. The exact prevalence of pediatric rare diseases is still unknown. Rare and orphan diseases include several syndromes, most of them with a genetic etiology. The diagnostic challenges to identify and confirm a rare disease are significant. The complexity of treating and managing a patient with a rare disease may be difficult for the general medical provider without adequate diagnostic tools and specialist referrals. An interdisciplinary approach for rare-disease management is critical to address symptoms and for patients to have access to effective medical treatments. A better understanding of the basic science, medical pathophysiology, and clinical diagnosis for current and future treatments is needed. Additionally, new strategies to involve caretakers in the community to participate in pediatric rare-disease research is important to understand rare etiologies of the disease, decrease medical barriers, improve the access to healthcare and avoid health disparities in this population.

children

IMPACT FACTOR 2.0 | Indexed in: PubMed | CITESCORE 2.7

an Open Access Journal by MDPI

Editor-in-Chief

Prof. Dr. Paul R. Carney
Departments of Child Health and Neurology, University of Missouri, 400 Keene Street, Columbia, MI 65201, USA

Message from the Editor-in-Chief

You are invited to contribute a research article or comprehensive review for consideration and publication in *Children* (ISSN 2227-9067). *Children* is an open access journal—research articles, reviews, and other content are published online immediately after acceptance. The scientific community and the general public have unlimited free access to the content as soon as it is published. The journal focuses on sharing clinical, epidemiological, and translational science relevant to children's health. We would be pleased to welcome you as one of our authors.

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, Embase, and other databases.
Journal Rank: JCR - Q2 (Pediatrics) / CiteScore - Q2 (Pediatrics, Perinatology and Child Health)

Contact Us

Children Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland

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

mdpi.com/journal/children
children@mdpi.com