Special Issue

Prevention and Detection of Pediatric Genetic Diseases

Message from the Guest Editor

Pediatric genetic diseases refer to conditions that are caused by genetic factors and affect children from birth or early childhood. These disorders, such as cystic fibrosis, sickle cell anemia, and autism spectrum disorder, can lead to life-limiting outcomes or lifelong care. While 1 in 20 children is affected by a rare genetic disorder, many conditions lack effective early detection methods. Current screening tests are limited, with many rare or complex diseases not included in standard newborn panels, and the genetic biomarkers for early diagnosis remain underexplored. There is also a scarcity of universal tests due to genetic heterogeneity and the complexity of multifactorial diseases. Newborn screening and prenatal tests such as NIPT are essential but are limited by their lack of sensitivity and specificity. Advanced genetic testing techniques such as WES and WGS offer promise but face challenges regarding their accessibility and data interpretation. Research is therefore needed to develop more effective screening biomarkers, enhance access to testing, and advance personalized therapies.

Guest Editor

Dr. Elham Khodayari Moez

L5-208, Mount Sinai Hospital, 60 Murray St., Toronto, ON M5T 3L9, Canada

Deadline for manuscript submissions

closed (10 July 2025)

Children

an Open Access Journal by MDPI

Impact Factor 2.1
CiteScore 3.8
Indexed in PubMed

mdpi.com/si/228011

Children
Editorial Office
MDPI, Grosspeteranlage 5
4052 Basel, Switzerland
Tel: +41 61 683 77 34
children@mdpi.com

mdpi.com/journal/children

Children

an Open Access Journal by MDPI

Impact Factor 2.1
CiteScore 3.8
Indexed in PubMed

mdpi.com/journal/children

MDPI

About the Journal

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.

Editor-in-Chief

Prof. Dr. Paul R. Carney
Departments of Child Health and Neurology, University of Missouri, 400 Keene Street, Columbia, MO 65211, USA

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)