Special Issue

Inborn Errors of Metabolism (IEMs): Advances in Diagnosis and Treatment

Message from the Guest Editor

Newborn screening for Inborn Errors of Metabolism (IEMs) is one the most important public health initiatives of this century. It was initially introduced in the 1960s as a part of the population screening for malignancies conducted by Dr. Robert Guthrie, who had previously realized that blood from a newborn baby's heel onto filter paper could be eluted and tested for phenylketonuria. Subsequently, other amino acid disorders, organic acidurias, and some urea cycle disorders could similarly be tested.
For several years, a single disorder was diagnosed through a single test. In the past, tandem mass spectrometry (MS-MS) was performed in order to test as many infants as possible in the newborn period. Currently, there is much discussion on whether it would be more beneficial to move to whole exome sequencing. However, there is also concern about cost and the interpretation of the results.

This Special Issue welcomes submissions of original research articles, review papers, and opinion pieces to share the latest findings, experiences, and perspectives on newborn screening and clinical management for inborn errors of metabolism (IEMs).

Guest Editor

Dr. Deborah Marsden
Department of Genetics, Children's Hospital Boston, 300 Longwood Ave, Boston, MA 02115, USA

Deadline for manuscript submissions

closed (25 July 2025)

GCAT
TACG
GCAT

Genes

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed

mdpi.com/si/229500

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

mdpi.com/journal/genes

MDPI

GCAT
TACG
GCAT

Genes

an Open Access Journal by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed

mdpi.com/journal/genes

MDPI

About the Journal

Message from the Editor-in-Chief

Genes is central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fast-moving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised. Why not consider *Genes* for your next genetics paper?

Editor-in-Chief

Prof. Dr. Selvarangan Ponnazhagan
Department of Pathology, The University of Alabama at Birmingham, 1825 University Blvd, SHEL 814, Birmingham, AL 35294-2182, 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, MEDLINE, PMC, Embase, PubAg, and other databases.

Journal Rank:

JCR - Q2 (Genetics and Heredity) / CiteScore - Q2 (Genetics (clinical))