

Special Issue

Genetic Research in Metabolic Diseases

Message from the Guest Editors

With more than 1,400 different genetic diseases affecting a variety of biochemical pathways, the field of metabolic diseases represents a dynamic and expanding field of genetic research. In recent years, the increased knowledge in genetics, has animated researchers and clinicians who are dealing with the complexity of inherited metabolic diseases (IMDs). In particular, the areas of lysosomal storage disorders (LSD), congenital disorders of glycosylation (CDG), and mitochondrial disorders have seen important advancements and discoveries. From newborn screening programs to gene therapy, these diseases offer a wide range of possibilities in genetic research, with the potential of an unprecedented impact on those affected. At the same time, the field is characterized by unique technical and ethical challenges.

This Special Issue on Genetic Research in Metabolic Diseases will provide an updated overview, novel insights, and critical perspectives in the pathophysiology, diagnosis, and treatment of inherited metabolic disorders such as LSD, CDG, and mitochondrial disorders. Contributions from experts in the field through research papers and reviews are welcome.

Guest Editors

Prof. Dr. Maurizio Scarpa

Regional Coordinating Center for Rare Diseases, European Reference Network for Hereditary Metabolic Diseases (MetabERN), Udine University Hospital, Piazzale Santa Maria della Misericordia 15, 33100 Udine, Italy

Prof. Dr. Peter Witters

Department of Pediatrics, Universitair Ziekenhuis Leuven, Leuven, Belgium

Deadline for manuscript submissions

closed (10 March 2023)

G C A T
T A C G
G C A T

Genes

an Open Access Journal
by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed



mdpi.com/si/81845

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

[mdpi.com/journal/
genes](https://mdpi.com/journal/genes)



G C A T
T A C G
G C A T

Genes

an Open Access Journal
by MDPI

Impact Factor 2.8
CiteScore 5.5
Indexed in PubMed



[mdpi.com/journal/
genes](https://mdpi.com/journal/genes)



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
Experimental Cancer Therapeutics, 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))