

Special Issue

Genetics and Metabolism in Skeletal Muscle Disorders

Message from the Guest Editors

In recent years, interest in skeletal muscle disorders has significantly increased. The metabolic myopathies related to skeletal muscle are a heterogeneous group of rare, genetically determined conditions characterised by defects in the biochemical pathways of storage, mobilisation and utilisation of the substrates useful for energy generation and the contraction of muscle cells. They can develop at any age and have varying clinical presentations. The aims of this Special Issue are to a) explore the developments in our understanding and treatment of these conditions; b) highlight new treatments available for skeletal muscle disorders, e.g., Pompe's disease. We welcome original research and brief case reports that focus on pathogenetic mechanisms; clinical features; and recent advances in the investigation and treatment of the principal metabolic muscle diseases, such as glycogen storage disorders (GSD), fatty acid oxidation disorders, disorders of purine metabolism and mitochondrial disorders. Research using cellular or animal models is also welcome.

Guest Editors

Dr. Michela Ripolone

Neuromuscular and Rare Disease Unit, ERN EURO-NMD, Paris, France

Dr. Simona Zanotti

Neuromuscular and Rare Disease Unit, ERN EURO-NMD, Paris, France

Deadline for manuscript submissions

closed (20 November 2022)



Metabolites

an Open Access Journal
by MDPI

Impact Factor 3.7
CiteScore 6.9
Indexed in PubMed



mdpi.com/si/123077

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

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





Metabolites

an Open Access Journal
by MDPI

Impact Factor 3.7
CiteScore 6.9
Indexed in PubMed



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



About the Journal

Message from the Editor-in-Chief

The metabolome is the result of the combined effects of genetic and environmental influences on metabolic processes. Metabolomic studies can provide a global view of metabolism and thereby improve our understanding of the underlying biology. Advances in metabolomic technologies have shown utility for elucidating mechanisms which underlie fundamental biological processes including disease pathology. *Metabolites* is proud to be part of the development of metabolomics and we look forward to working with many of you to publish high quality metabolomic studies.

Editor-in-Chief

Dr. Amedeo Lonardo

Internal Medicine, Ospedale Civile di Baggiovara, Azienda Ospedaliero-Universitaria, 41126 Modena, Italy

Author Benefits

High Visibility:

indexed within Scopus, SCIE (Web of Science), PubMed, PMC, Embase, CAPlus / SciFinder, and other databases.

Journal Rank:

JCR - Q2 (Biochemistry and Molecular Biology) / CiteScore - Q2 (Endocrinology, Diabetes and Metabolism)

Rapid Publication:

manuscripts are peer-reviewed and a first decision is provided to authors approximately 14.4 days after submission; acceptance to publication is undertaken in 3.6 days (median values for papers published in this journal in the first half of 2025).