



Diagnosis and Management of Lysosomal Storage Diseases in Specific Body Organs and Systems

Guest Editors:

Dr. Patryk Lipiński

Institute of Clinical Sciences,
Maria Skłodowska-Curie Medical
Academy, Warsaw, Poland

**Prof. Dr. Anna Tyłki-
Szymańska**

Department of Pediatrics,
Nutrition and Metabolic Diseases,
The Children's Memorial Health
Institute, Al. Dzieci Polskich 20,
04-730 Warsaw, Poland

Deadline for manuscript
submissions:

30 November 2024

Message from the Guest Editors

Dear Colleagues,

Lysosomal storage diseases (LSDs) are a group of monogenic metabolic disorders associated with the dysfunction of lysosomal apparatus. Virtually every cell in the body possesses lysosomes, yet storage in these organelles can vary even among various cells. The clinical phenotype is quite heterogeneous, affecting multiple organs and systems, reflecting also the complexity of their pathomechanism. Even though our knowledge of LSD has increased, and many therapies have been established, many aspects of their presentation remain ambiguous.

The aim of this Special Issue is to provide a comprehensive overview of clinical presentation, diagnostics and treatment, focusing on specific body organs and systems. Potential topics include, but are not limited to, the following:

1. Liver in LSD;
2. Cardiac involvement in LSD;
3. Skeletal complications in LSD;
4. Respiratory system involvement in LSD;
5. Central and peripheral nervous systems abnormalities in LSD;
6. Biomarkers in LSD diagnostics;
7. Genetics, inheritance, founder effects and genotype-phenotype correlation.





an Open Access Journal by MDPI

Editor-in-Chief

Prof. Dr. Andreas Kjaer

Department of Clinical
Physiology, Nuclear Medicine &
PET National University Hospital,
Rigshospitalet, University of
Copenhagen, Blegdamsvej 9, DK-
2100 Copenhagen, Denmark

Message from the Editor-in-Chief

You are cordially invited to submit research articles, short communications, comprehensive reviews, case reports or interesting images for consideration and publication in *Diagnostics* (ISSN 2075-4418). *Diagnostics* is published in open access format – research articles, reviews and other contents are released on the Internet immediately after acceptance. The scientific community and the general public have unlimited and free access to the content as soon as it is published. 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, Inspec, CAPlus / SciFinder, and other databases.

Journal Rank: JCR - Q1 (Medicine, General and Internal) / CiteScore - Q2 (Internal Medicine)

Contact Us

Diagnostics Editorial Office
MDPI, St. Alban-Anlage 66
4052 Basel, Switzerland

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

mdpi.com/journal/diagnostics
diagnostics@mdpi.com
[X@diagnostic_mdpi](https://twitter.com/diagnostic_mdpi)