

## Special Issue

# Newborn Screening for Lysosomal Storage Disorders

### Message from the Guest Editors

Newborn screening for lysosomal storage disorders (LSDs) has gained significant momentum during the past two years, stimulated by the addition of Pompe Disease and Hurler Syndrome to the recommended universal screening panel (RUSP). New therapies, especially enzyme replacement and hematopoietic stem cell transplantation, have given new hope to families with affected children. Also, the development of fluorometric enzymatic assays for LSDs in dried blood pioneered by Nestor Chamoles in 2001 and boosted by Michael Gelb's tandem mass spectrometry method (2004) enables early detection of newborns. Multiplexing of LSD enzyme assays using either MS/MS or digital microfluidic fluorometry has facilitated high-throughput prospective screening for multiple LSDs. Many have argued against screening for LSDs, citing the expense and variable efficacy of current treatment options and the preponderance of cases with later onset forms of disease, pseudodeficiency alleles, genetic variants of unknown significance and carriers. Post-analytical methods including statistical analytical tools and second-tier biochemical tests should help clarify these challenges.

---

### Guest Editors

Prof. Dr. David S. Millington

Professor Emeritus of Pediatrics, Medical Genetics Division, Duke Medicine, Durham, NC 27709, USA

Dr. Janice Fletcher

Genetics and Molecular Pathology, SA Pathology at Women's and Children's Hospital, 72 King William Road, North Adelaide, SA 5006, Australia

---

### Deadline for manuscript submissions

closed (30 September 2018)



## International Journal of Neonatal Screening

---

an Open Access Journal  
Published by MDPI

---

Impact Factor 4.0  
CiteScore 6.0  
Indexed in PubMed



[mdpi.com/si/12492](https://mdpi.com/si/12492)

*International Journal of  
Neonatal Screening*  
Editorial Office  
MDPI, Grosspeteranlage 5  
4052 Basel, Switzerland  
Tel: +41 61 683 77 34  
[ijns@mdpi.com](mailto:ijns@mdpi.com)

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





# International Journal of Neonatal Screening

---

an Open Access Journal  
Published by MDPI

---

Impact Factor 4.0  
CiteScore 6.0  
Indexed in PubMed



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



## About the Journal

### Message from the Editorial Board

---

#### Editors-in-Chief

Dr. Peter C. J. I. Schielen

Office of the International Society for Neonatal Screening, Reigerskamp  
273, 3607 HP Maarssen, The Netherlands

Dr. Ralph Fingerhut

Newborn Screening and Metabolic Laboratory, Synlab MVZ Weiden, D-  
92637 Weiden, Germany

---

#### Author Benefits

##### High Visibility:

indexed within Scopus, ESCI (Web of Science), PubMed,  
PMC, Embase, and other databases.

##### Journal Rank:

JCR - Q1 (Pediatrics) / CiteScore - Q1 (Pediatrics,  
Perinatology and Child Health)

##### Rapid Publication:

manuscripts are peer-reviewed and a first decision is  
provided to authors approximately 26.1 days after  
submission; acceptance to publication is undertaken in 3.9  
days (median values for papers published in this journal in  
the first half of 2025).