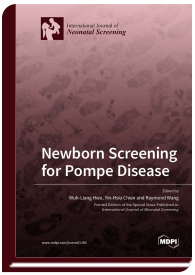
International Journal of
Neonatal Screening

IMPACT FACTOR 3.5
Indexed in: PubMed
CITESCORE 5.0

Special Issue Reprint

Newborn Screening for Pompe Disease



www.mdpi.com/books/reprint/4026

Edited by
Wuh-Liang Hwu
Yin-Hsiu Chien
Raymond Wang

ISBN 978-3-0365-0580-0 (Hardback)
ISBN 978-3-0365-0581-7 (PDF)

Pompe disease, also known as acid maltase deficiency or acid alpha-glucosidase deficiency, in its most severe form results in a rapidly progressive, neonatal-onset skeletal and cardiomyopathy, leading to early infantile death without treatment. The development of treatment with recombinant enzyme replacement therapy radically transformed the clinical trajectory of those affected, enabling long-term ventilator-free survival with resolution of cardiomyopathy. These positive clinical outcomes resulted in the implementation of newborn screening programs for Pompe disease across the world. This Special Issue highlights some of the experiences of Pompe screening programs worldwide and discusses public policy and ethical issues elicited by presymptomatic screening for Pompe disease.

MDPI

Order Your Print Copy
You can order print copies at
www.mdpi.com/books/reprint/4026

MDPI Books

Publishing Open Access Books & Series

MDPI Books offers quality open access book publishing to promote the exchange of ideas and knowledge in a globalized world. MDPI Books encompasses all the benefits of open access – high availability and visibility, as well as wide and rapid dissemination. With MDPI Books, you can complement the digital version of your work with a high quality printed counterpart.

Open Access

Your scholarly work is accessible worldwide without any restrictions. All authors retain the copyright for their work distributed under the terms of the Creative Commons Attribution License.

Author Focus

Authors and editors profit from MDPI's over two decades of experience in open access publishing, our customized personal support throughout the entire publication process, and competitive processing charges as well as unique contributor discounts on book purchases.

High Quality & Rapid Publication

MDPI ensures a thorough review for all published items and provides a fast publication procedure. State-of-the-art research and time-sensitive topics are released with a minimum amount of delay.

High Visibility

Due to our global network and well-known channel partners, we ensure maximum visibility and broad dissemination. Title information of books is sent to international indexing databases and archives, such as the Directory of Open Access Books (DOAB), and the Verzeichnis Lieferbarer Bücher (VLB).

Print on Demand and Multiple Formats

MDPI Books are available for purchase and to read online at any time. Our print-on-demand service offers a sustainable, cost-effective and fast way to publish MDPI Books printed versions.

MDPI AG
St. Alban-Anlage 66
4052 Basel
Switzerland
Tel: +41 61 683 77 34
www.mdpi.com/books
books@mdpi.com