

Lipid Storage

including sphingomyelin and cholesterol accumulation



SMPD1 gene mutation cause acid sphingomyelinase activity deficiency



Niemann-Pick disease type A and B

Glycolipid Storage

Glucocerebrosidase deficiency



Gaucher's disease

Glycosphingolipid Storage

α-galactosidase A deficiency



Fabry diseases

Mutations of NPC1 and NPC2 genes cause impairment in cellular processing and transport of LDL-cholesterol



Niemann-Pick disease type C

Glycogen Storage

Acid alpha-glucosidase or acid maltase deficiency

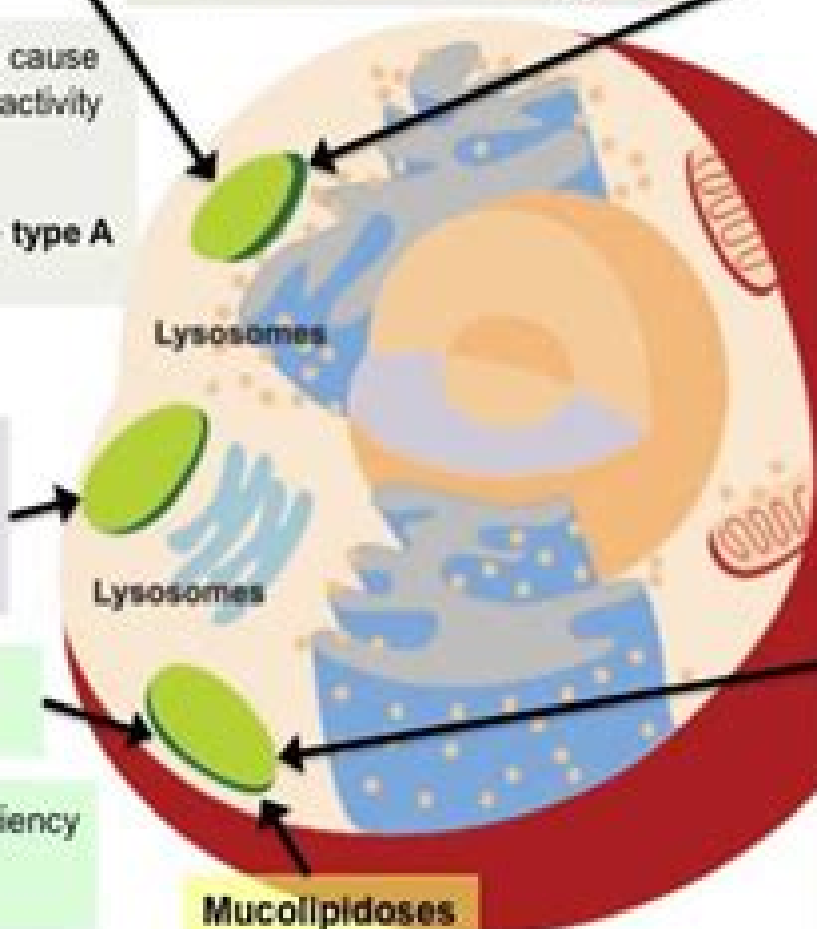


Pompe disease

Glycosaminoglycans (mucopolysaccharides) Storage

- α-L-iduronidase deficiency → mucopolysaccharidosis I
- Iduronate sulfatase deficiency → mucopolysaccharidosis II
- Heparan sulfamidase deficiency → mucopolysaccharidosis IIIA
- N-acetylglucosaminidase deficiency → mucopolysaccharidosis IIIB
- Heparan-α-glucosaminide N-acetyltransferase deficiency → mucopolysaccharidosis IIIC
- N-acetylglucosamine 6-sulfatase deficiency → mucopolysaccharidosis IIID
- Galactose-6-sulfate sulfatase deficiency → mucopolysaccharidosis IVA
- β-galactosidase deficiency → mucopolysaccharidosis IVB
- N-acetylgalactosamine-4-sulfatase deficiency → mucopolysaccharidosis VI
- β-glucuronidase deficiency → mucopolysaccharidosis VII
- Hyaluronidase deficiency → mucopolysaccharidosis IX

- α-N-acetyl neuraminidase (sialidase) → mucopolipidosis type I
- N-acetylglucosamine-1-phosphotransferase deficiency → mucopolipidosis type II and type III
- MCONLN1 gene mutation encoding mucopolipin1 → mucopolipidosis type IV



Lysosomal Storage Disease

Gregory M. Pastores



Lysosomal Storage Disease:

Lysosomal Storage Disorders Atul B. Mehta, Bryan Winchester, 2022-07-12 *Lysosomal Storage Disorders* Enables readers to gain both holistic and specific knowledge on the topic of Lysosomal Storage Disorders In the past few decades we have witnessed a golden age of Lysosomal Storage Disorders LSDs clinical care diagnosis and research and this book is a well timed review of the rapidly developing subject of LSDs Contributions from many leading scientists and clinicians in the field provide the reader with a completely comprehensive overview of the subject Topics covered in the book include The general aspects of LSDs with special attention paid to physiology and pathology Clinical and laboratory diagnosis including newborn screening and the genetics of LSDs Individual LSDs such as the various sphingolipidoses mucopolysaccharidoses glycogen storage disease glycoproteinoses galactosialidosis and neuronal ceroid lipofuscinoses Other disorders of the lysosome including those involving defects in the lysosomal membrane and in the emerging roles of the lysosome in cellular metabolism The existing and emerging treatments for LSDs Key patient issues such as availability and disease awareness including in the Third World For researchers and industry professionals in the field of LSDs this book serves as a completely comprehensive reference work to understand both theoretical concepts and how they may be used in practical applications For students of LSDs it provides a solid base of foundational knowledge and serves as a practical guide *Lysosomal Storage Disorders* John A. Barranger, Mario Cabrera-Salazar, 2007-10-16 Scientific progress has been rapid in lysosomal biology during the last six decades Its application to human disease is nothing less than spectacular In no other group of disorders has knowledge and clinical utility progressed so speedily Recall that the organelle was described in just 1955 Since then the biochemical alterations and storage materials were described the enzyme deficiencies discovered the gene coding of these glycoproteins cloned and thousands of mutations defined These advances have resulted in highly improved diagnosis for more than 50 diseases For five diseases including the most common lysosomal storage disorder molecular therapy is a reality extremely effective and very safe This higher plateau of medical approaches to human disease is something to which all translational scientists aspire and only a few actually witness The relief of pain and suffering is a tribute to the ideas and work of many dedicated investigators Much of that work is presented in this text Despite our ability to treat some of these diseases through enzyme replacement therapy ERT and accurately define different diseases that look alike there is much to be learned about lysosomal disease With each step up the barrier to knowledge a new point of view is attained a fresh perspective Much is seen more clearly and many allegories of the cave are dispelled forever Yet our new view demands a new vision drawing us to find better definitions of what we see This is how it has been with lysosomal diseases *Lysosomal Storage Disorders: Principles And Practice* Gregory M Pastores, 2009-12-14 This book presents an overview of lysosomal storage disorders and provides the reader with an understanding of clinical features associated complications and diagnosis and management approaches It also describes historical developments in the field and current thinking relating to

pathophysiology and prospective therapeutic strategies The book is written by an expert in the field who has been engaged in both basic and clinical research in addition to having extensive practical experience in patient care It is written from the perspective of someone who entered the field just as treatment was being introduced and who has been engaged in the seminal clinical trials and the development of therapeutic guidelines It offers a broad perspective and should appeal to both novices and experts in the field who seek a single resource that provides a comprehensive picture of relevant topics on this subject A multi faceted volume the author addresses the issue of diagnosis and patient management underlying mechanisms of disease sources of morbidity and treatment options covering issues of interest to both the basic scientist and the clinician

Lysosomes and Lysosomal Storage Diseases John W. Callahan,J. Alexander Lowden,1981 **Lysosomal Storage**

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Pastores,2016 **Lysosomal Storage Disorders** John A. Barranger,Mario Cabrera-Salazar,2007-10-01 The knowledge of lysosomal biology and the consequences of its dysfunction have increased dramatically in the past 60 years This book describes the nature of the lysosomal dysfunction and diseases as well as potential future treatments and therapies Disease specific chapters provide thorough reviews of the clinical features of lysosomal storage disorders their molecular basis and the commercial or experimental therapeutic approaches sought in this area This is an invaluable resource for researchers in biochemical and molecular genetics enzyme therapy and gene transfer **Lysosomal Storage Diseases** R. W. E.

Watts,Dorothy A. Gibbs,1986 **Molecular Basis of Lysosomal Storage Disorders** John Barranger,2012-12-02 Molecular Basis of Lysosomal Storage Disorders contains the proceedings of the 1983 Conference on the Molecular Basis of Lysosomal Storage Disorders held at the National Institutes of Health in Bethesda Maryland The papers focus on the molecular biology of and therapeutic approaches to lysosomal storage disorders such as mucopolysaccharidoses sphingolipidoses and Gaucher disease Organized into six sections comprised of 29 chapters this book begins with an overview of enzymes activator proteins and stabilizers that underlie lysosomal storage disorders It then discusses some developments in enzyme purification

receptors for glycoprotein enzymes factors that control endocytosis and the intracellular fate of lysosomal hydrolases Some chapters explain the enzyme biosynthesis bone marrow transplantation and enzyme replacement along with cell hybridization chromosome localization phenotype discrimination and cloning of genes for human lysosomal enzymes This book is helpful to biochemists physiologists pathologists geneticists clinical investigators and practicing physicians concerned with the study care and treatment of patients with hereditary metabolic disorders as well as undergraduate and graduate level students involved in research in this discipline *Lysosomal Storage Disorders* Enrico Moro, 2021-11-12

Lysosomal storage disorders are a heterogeneous group of rare genetic conditions affecting worldwide population and often exhibiting severe clinical manifestations During the last two decades the joined collaboration between scientists and clinicians has allowed to offer valuable therapeutic options to affected patients Therefore the tight connection between basic science and clinical medicine represents the gold standard approach to these disorders In this context the present book collects a piece of current scientific advances in the knowledge of disease pathogenesis and in the development of novel diagnostic and therapeutic strategies for some of these diseases Altogether these articles define and recapitulate which essential steps are required during the clinical management of a rare inherited disorder and describe forthcoming advances and a breakthrough in the field of lysosomal diseases

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