

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

- α-N -acetyl neuraminidase (sialidase) → mucopolipidosis type I
- N-acetylglucosamine-1- phosphotransferase deficiency → mucopolipidosis type II and type III
- MCONLN1 gene mutation encoding mucolin1 → mucopolipidosis type IV

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



Niemann-Pick disease type C

Glycogen Storage

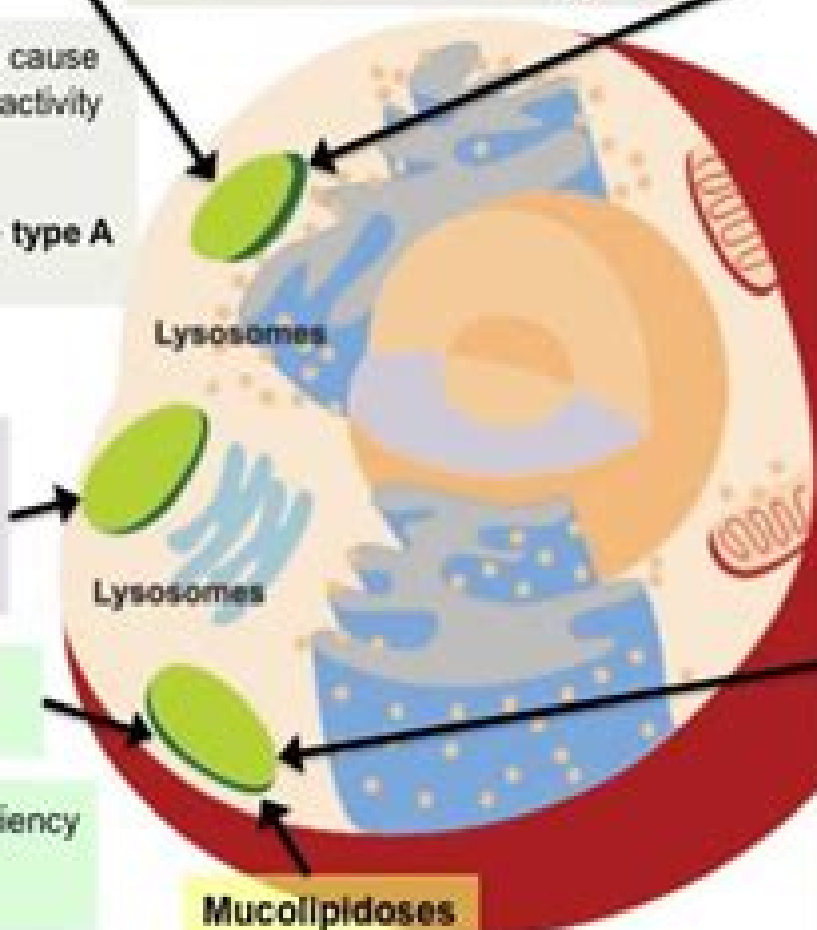
Acid alpha-glucosidase or acid maltase deficiency



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



Lysosomes And Storage Diseases

Gregory M. Pastores



Lysosomes And Storage Diseases:

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 Lysosomes and Storage Diseases Henri-Géry Hers, François Van Hoof, 1973 *Lysosomes and Lysosomal Storage Diseases* John W. Callahan, J. Alexander Lowden, 1981 **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 **Lysosomal Storage Diseases** Gregory M. Pastores, 2016 Lysosomal Storage Diseases Gregory M. Pastores, 2016 This monograph presents an overview of the lysosome and its different roles in the maintenance of cellular homeostasis It also provides a description of the various pathophysiologic changes associated with the lysosomal storage disorders Therapeutic considerations and aspects relating to genetic counselling are covered in the final section Several diagrams are provided to facilitate greater understanding of

relevant themes Additionally contextual material is given at the end of each chapter which elaborates on some of the entries brought up in the discussions The book is written by an expert in the field who has been engaged in basic and clinical research 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 examination of the relevant topics Issues covered should appeal to both the basic scientist and the clinician

Lysosomal Storage Disorders Atul B. Mehta, Bryan Winchester, 2012-09-06 The last two decades have seen a huge expansion in research in the area of lysosomal storage disorders which has substantially extended our understanding of both the scientific and the clinical basis of these diseases Lysosomal Storage Disorders A Practical Guide is the fruit of an ambitious project aiming to review both the scientific and the clinical aspects of lysosomal storage disorders resulting in this accessible volume which gives an up to date overview of the subject There is substantial scientific interest in these diseases new advances in small molecule therapy are likely to be useful in the near future and trials are already underway Lysosomal storage disorders offer a unique platform for teaching modern clinical science from basic genetics through to clinical applications The first part of the book reviews and classifies our current understanding of the physiology and pathophysiology of lysosomal storage disorders The second part of the book reviews individual diseases and gives perspectives from patients and experts looking towards future therapeutic directions Lysosomal Storage Disorders A Practical Guide is the ideal guide for a wide audience including scientists clinicians health care workers and administrators those working in the pharmaceutical industry patients and their organisations Titles of related interest Haematology at a Glance Mehta ISBN 9781405179706 Atlas of Endocrine and Metabolic Disease Pozzilli ISBN 9780470656273 [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 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. *Lysosomes* Paul Saftig, 2007-03-20

Lysosomes are membrane surrounded organelles which are present in all animal cells. The importance of this organelle is underlined by an increasing number of human diseases which are associated with an impaired function of the lysosomal compartment. This book summarizes the current state of the art knowledge about this unique organelle. It addresses the biogenesis of this compartment, the transport of lysosomal proteins, the role of the lysosomal membrane in lysosomal stability and transport, the function of lysosomal proteases and hydrolases, lysosomal storage disorders and new concepts on how to treat these diseases. In addition to these classical topics, new insights into lysosomal functions are covered by chapters dealing with specialized lysosomes involved in bone resorption and plasma membrane repair, the lysosomal transcriptome and proteome and the emerging role of lysosomes in special forms of autophagy. This book will provide readers with a comprehensive overview into how this fascinating organelle works and how research in the field is developing.

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