

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

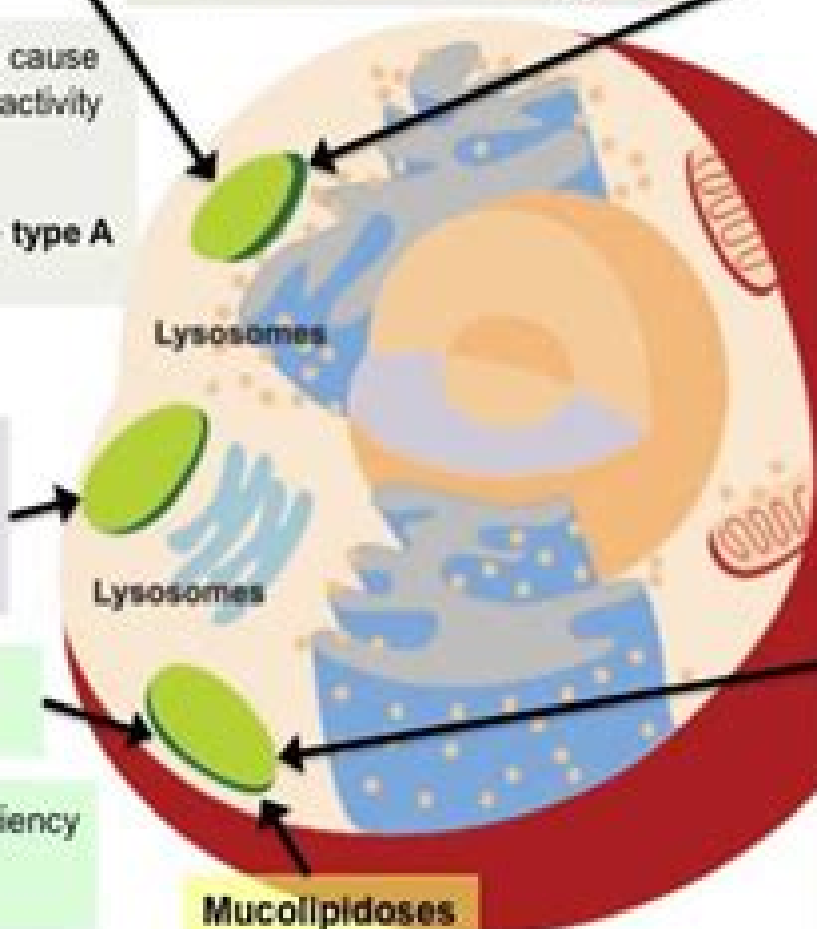
Acid alpha-glucosidase or acid maltase deficiency



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

Lysosomes

Mucopolipidoses

Lysosomes And Lysosomal Storage Diseases

**Frederick R. Maxfield, James M.
Willard, Shuyan Lu**



Lysosomes And Lysosomal Storage Diseases:

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

Lysosomes and Lysosomal Diseases , 2015-02-04 This new volume of *Methods in Cell Biology* looks at methods for lysosomes and lysosomal diseases Chapters focus upon practical experimental protocols to guide researchers through the analysis of multiple aspects of lysosome biology and function In addition it details protocols relevant to clinical monitoring of patients with lysosomal diseases With cutting edge material this comprehensive collection is intended to guide researchers for years to come Covers sections on model systems and functional studies imaging based approaches and emerging studies Chapters are written by experts in the field Cutting edge material *Lysosomal Storage Diseases* Gregory M. Pastores, 2016

Lysosomes Frederick R. Maxfield, James M. Willard, Shuyan Lu, 2016-06-27 Discussing recent findings up to date research and novel strategies the book integrates perspectives from pharmacology toxicology and biochemistry to illustrate the potential of lysosomes in drug discovery and development Explores basic principles and properties of lysosomes that allow them to act as regulators of cell metabolism therapeutic targets and sites for activation of drug conjugates Discusses the role of lysosomes in metabolism drug targeting apoptosis cancer aging inflammation autophagy metabolism toxicity and membrane repair Introduces new pathways in therapeutic development and new mechanisms in drug development

Lysosomal Storage Diseases R. W. E. Watts, Dorothy A. Gibbs, 1986 *Lysosomes and Storage Diseases* Henri-Géry Hers, François Van Hoof, 1973 *Inherited Metabolic Epilepsies* Phillip L. Pearl, 2012-10-04 The explosion of information in neurogenetics and metabolism mandates increasing awareness of appropriate diagnostic and therapeutic strategies in the setting of certain epilepsies especially those of very early onset There are over 200 inherited disorders that are associated with seizures and prompt identification and intervention is crucial for a positive outcome This text brings together the leading authorities working in this area to present state of the art clinical reviews covering the science recognition and treatment of the inherited metabolic epilepsies and related disorders The book begins with general principles for diagnosis

and targeted intervention including screening protocols laboratory testing seizure patterns and EEG findings imaging new technologies and the ketogenic diet The next two sections are devoted to the cohort of specific small molecule and large molecule disorders that are treatable yet can be so vexing to clinicians and investigators The book concludes with a clinical algorithm designed to be a resource for the physician in search of direction while considering an inherited metabolic disorder as the explanation for a patient with epilepsy

Clinical Neuropathology R. O. Weller, M. Swash, D. L. McLellan, C. L. Scholtz, 2012-12-06 Although most textbooks of neurology contain a certain amount of pathological information neuropathology has often been treated in isolation However neuropathology has a close relationship to clinical neurology neurosurgery and neuroradiology Thus advances in the rapidity and accuracy of pathological diagnosis have often led to changes in clinical management and recently improvements in clinical diagnosis particularly CT scanning have brought about a change in emphasis in the practice of neuropathology In this textbook we have sought to present a widely based account of neuropathology in combination with information from clinical experience We chose this approach in order to emphasize the close interrelation between clinician and pathologist The book grew out of a course organised jointly by two neurologists and two neuropathologists from the Departments of Neuropathology and Neurology of The London Hospital and The University of Southampton It is hoped that the book will be useful not only to pathologists neurologists neurosurgeons and neuroradiologists but also to general physicians In a period of rapid advance in knowledge it is important to recognise how changes in the clinical and laboratory disciplines overlap In order to make the most of consultations with pathologist colleagues the clinician must know what skills and techniques are available in the laboratory and similarly the pathologist must keep abreast of changes in clinical practice In the past the clinician and pathologist have often been slow to appreciate advances in each other's fields

Biology of the Lysosome John B. Lloyd, Robert W. Mason, 2012-12-06 Volume 27 provides a comprehensive review of current knowledge of lysosome function in mammalian cells The book's unique contribution is its series of chapters that offers unparalleled treatment of the metabolic activities of lysosomes

Adopting the Melody of Appearance: An Mental Symphony within **Lysosomes And Lysosomal Storage Diseases**

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