

Lysosomal Storage Diseases

S.N	Disease Name	Substrate	Enzyme/Protein	Gene
1	Tay-sachs disease	Ganglioside-gm2	Beta-hexosaminidase A	HEXA
2	Gaucher disease	Glucocerebroside	Beta-glucocerebrosidase	GBA
3	Hunter syndrome	Sulfated alpha-l-iduronic acid	Iduronate 2-sulfatase	IDS
4	Pompe disease	Glycogen	Acid maltase	GAA
5	Niemann pick disease	Sphingomyelin	Acid sphingomyelinase	SMPD1
6	Fabry disease	Globotriaosylceramide	Alpha-galactosidase A	GLA
7	Metachromatic leukodystrophy	Sulfatides	Arylsulfatase A	ARSA
8	Krabbe	Galactosylceramide	Galactosylceramidase	GALC
9	Salla disease	Sialic acid	Sialin	SLC17A5
10	Obalamin C disease	Cobalamin	Cobalamin reductases	MMACHC
11	Mucopolidosis II	Functional components	Glcnac-1-phosphotransferase	GNPTAB
12	Sandhoff disease	GM2 ganglioside	Beta-hexosaminidase	HEXB
13	Cystinosis	Cystine	Cystinosis	CTNS

Lysosomes And Lysosomal Storage Diseases

Leslie E. Silberstein, John Anastasi



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 *Hematology: Basic Principles and Practice E-Book* Leslie E. Silberstein, John Anastasi, 2017-06-14 Get the expert guidance you need to offer your patients the best possible outcomes with Hematology Basic Principles and Practice 7th Edition This thoroughly up to date text contains both unparalleled scientific content and must know clinical guidance so you can enhance your problem solving skills and make optimal use of the newest diagnostic techniques and therapeutic options in this fast changing field Delivers state of the art information and guidance from editors and global contributors who are at the forefront of their respective subspecialty areas Features sweeping content updates

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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

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