# lysosomal disorders of the brain

F. M. PLATT | S. U. WALKLEY

# <u>Lysosomal Disorders Of Brain Recent Advances In</u> <u>Molecular And Cellular Pathogenesis And Treatment</u>

**David Baud** 

#### Lysosomal Disorders Of Brain Recent Advances In Molecular And Cellular Pathogenesis And Treatment:

Lysosomal Disorders of the Brain ,2004 Lysosomal Disorders of the Brain Steven U. Walkley, 2004 Lysosomal storage diseases are inherited metabolic disorders characterized by severe pathology typically involving the brain Although individually rare they collectively represent a significant group of diseases that primarily present in early infancy or childhood In recent years considerable progress has been made in understanding the molecular mechanisms that lead to disordered function of the lysosomal system and to lysosomal storage Unravelling the basis for these diseases is providing unique insight into the normal biology of cells and pointing the way to the development of therapeutic strategies for their treatment Lysosomal Disorders of Brain details recent advances in the molecular and cellular pathologies of these diseases and in the development of effective therapies After an overview of the biology of the endosomal lysosomal system and the types of diseases resulting from defects in this system the book describes in detail the molecular mechanisms of storage model systems and pathophysiological mechanisms and finally new advances toward treatment With each chapter written by leading experts in their field this book will be valuable for scientists and clinicians in helping them understand the role of lysosomes in normal cells and mechanisms underlying these disorders how they can be diagnosed and the treatment options that are currently available Lysosomal Storage Disorders Atul B. Mehta, Bryan Winchester, 2022-07-11 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 Lysosomes Frederick R. Maxfield, James M. Willard, Shuyan Lu, 2016-05-31 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

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metabolism and its role in metabolic diseases Focusing on the sphingolipid de novo synthesis pathway the effect of sphingomyelin ceramide and sphingosine 1 phosphate and linkage between sphingolipids and other lipids such as cholesterol it covers serine palmitoyltransferase ceramide synthases ceramidases sphingosine kinases and sphingomyelin synthases and more While highlighting how rare diseases related to abnormal glycosphingolipid metabolism this publication introduces sphingolipid metabolism related diseases such as lung diseases and cancers as well as sphingolipid circadian regulation The book demonstrates advances and limitations of research on sphingolipid metabolism and its roles in metabolic diseases and other diseases It offers graduate students and researchers a coherent overview of sphingolipids as well as the limitations of current research in the field and promotes further studies on metabolic diseases as well as pharmaceutical research on drug discovery based on sphingolipid de novo synthase Departments of Labor, Health and Human Services, Education, and Related Agencies Appropriations for 2006: National Institutes of Health United States. Congress. House. Committee on Appropriations. Subcommittee on the Departments of Labor, Health and Human Services, Education, and **Encyclopedia of Cell Biology**, 2015-08-07 The Encyclopedia of Cell Biology Four Volume Set Related Agencies, 2005 offers a broad overview of cell biology offering reputable foundational content for researchers and students across the biological and medical sciences This important work includes 285 articles from domain experts covering every aspect of cell biology with fully annotated figures abundant illustrations videos and references for further reading Each entry is built with a layered approach to the content providing basic information for those new to the area and more detailed material for the more experienced researcher With authored contributions by experts in the field the Encyclopedia of Cell Biology provides a fully cross referenced one stop resource for students researchers and teaching faculty across the biological and medical sciences Fully annotated color images and videos for full comprehension of concepts with layered content for readers from different levels of experience Includes information on cytokinesis cell biology cell mechanics cytoskeleton dynamics stem cells prokaryotic cell biology RNA biology aging cell growth cell Injury and more In depth linking to Academic Press Elsevier content and additional links to outside websites and resources for further reading A one stop resource for students researchers and teaching faculty across the biological and medical sciences Neurobiology of Disease ,2011-09-06 Neurobiology of Disease is aimed at any basic scientist or clinician scientist teaching a course or conducting research on the basic science underlying the major neurological diseases It provides an excellent overview of cutting edge research on the fundamental disorders of the nervous system including physiological and molecular aspects of dysfunction The major categories of neurological disease are covered and the chapters provide specific information about particular diseases exemplifying each of these categories Sufficient clinical information is included to put into perspective the basic mechanisms discussed The book assembles a world class team of section editors and chapters written by acknowledged experts in their respective fields Provides cutting edge information about fundamental mechanisms underlying neurological diseases Amply

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