

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

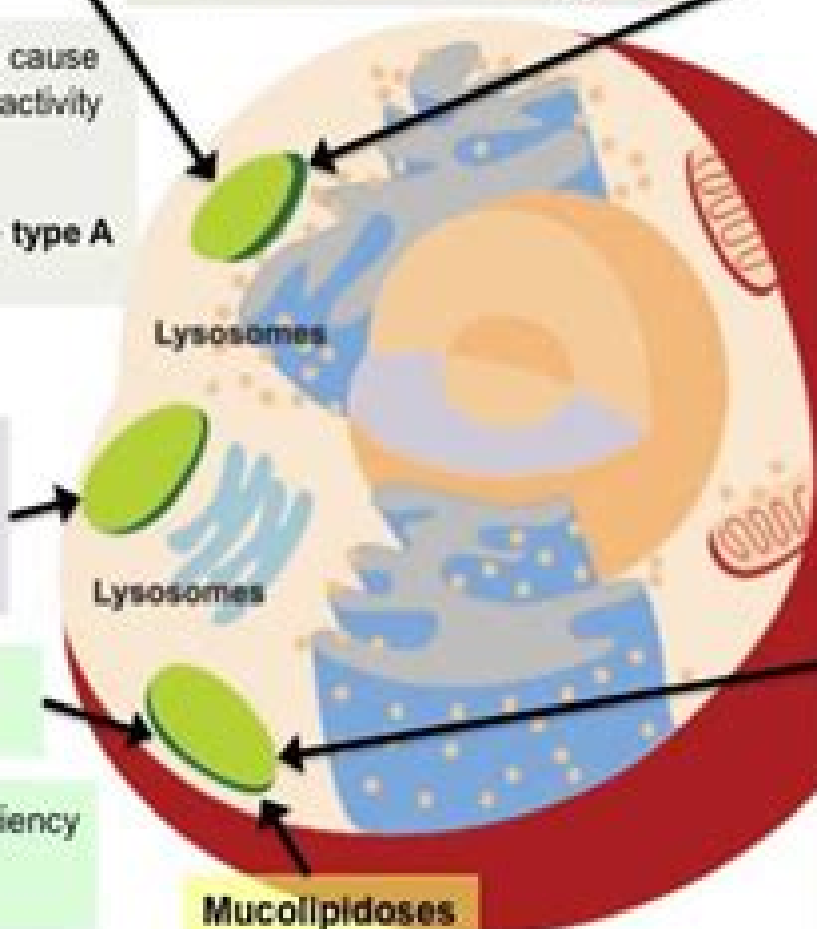


Pompe disease

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 Aspartylglycosaminuria

Ilkka Mononen



Lysosomal Storage Disease Aspartylglycosaminuria:

Lysosomal Storage Disease Ilkka Mononen,Nathan N. Aronson,1997 **Lysosomal Storage Disease** Ilkka Mononen,Nathan N. Aronson,1997-01-01 This volumes provides a comprehensive and up to date review of aspartylglycosaminuria AGU which is one of the most common lysosomal diseases worldwide It gives an overview of the disease from its discovery and early history to the most recent developments It also provides detailed reference material for clinical specialists and researchers working in the field of inborn errors of metabolism in particular on lysosomal diseases

Lysosomal Storage Diseases Parvesh K. Masson,1975 Greenfield's Neuropathology, 2-Volume Set, Eighth Edition Seth Love,David Louis,David W Ellison,2008-02-29 Greenfield s Neuropathology the worlds leading neuropathology reference provides an authoritative comprehensive account of the pathological findings in neurological disease their biological basis and their clinical manifestations This account is underpinned throughout by a clear description of the molecular and cellular processes and reactions that are relevant to the development and normal and abnormal functioning of the nervous system While this scientific content is of paramount importance however care has been taken to ensure that the information is presented in a way that is accessible to readers working within a range of disciplines in the clinical neurosciences and that also places the neuropathological findings within the context of a broader diagnostic process The new eighth edition incorporates much new information new illustrations and many new authors while retaining the depth breadth and quality of content so praised in previous editions Each chapter opens with an introductory section designed to offer an integrated approach to diagnosis taking account of clinical manifestations neuroradiological and laboratory findings as well as the neuropathological and molecular genetic features of the diseases being considered Strong emphasis has been placed on facilitating the retrieval of neuropathological information by non neuropathologists grappling with differential diagnoses or seeking information on broad categories of neurological disease and boxes and tables are used to present important symptoms and signs patterns of disease and other features for ease of reference High quality line and photographic illustrations the majority in full colour are all available on a companion CD to complete the offering *Greenfield's Neuropathology Eighth Edition 2-Volume Set* Seth Love,David Louis,David W Ellison,2008-02-29 Greenfield s Neuropathology the worlds leading neuropathology reference provides an authoritative comprehensive account of the pathological findings in neurological disease their biological basis and their clinical manifestations This account is underpinned throughout by a clear description of the molecular and cellular processes and reactions that are relevant to the development and normal and abnormal functioning of the nervous system While this scientific content is of paramount importance however care has been taken to ensure that the information is presented in a way that is accessible to readers working within a range of disciplines in the clinical neurosciences and that also places the neuropathological findings within the context of a broader diagnostic process The new eighth edition incorporates much new information new illustrations and many new authors while retaining

the depth breadth and quality of content so praised in previous editions Each chapter opens with an introductory section designed to offer an integrated approach to diagnosis taking account of clinical manifestations neuroradiological and laboratory findings as well as the neuropathological and molecular genetic features of the diseases being considered Strong emphasis has been placed on facilitating the retrieval of neuropathological information by non neuropathologists grappling with differential diagnoses or seeking information on broad categories of neurological disease and boxes and tables are used to present important symptoms and signs patterns of disease and other features for ease of reference High quality line and photographic illustrations the majority in full colour are all available on a companion CD to complete the offering

Greenfield's Neuropathology - Two Volume Set Seth Love,Arie Perry,James Ironside,Herbert Budka,2018-10-08
Greenfield's Neuropathology the world's leading neuropathology reference provides a comprehensive account of the pathological findings in neurological disease their biological basis and their clinical manifestations The book's detailed advice on pathological assessment and interpretation is based on clear descriptions of molecular and cellular processes and reactions that are relevant to the development of the nervous system as well as its normal and abnormal functioning The information is presented in an accessible way to readers working within a range of disciplines in the clinical neurosciences and neuropathological findings are placed within the context of a broader diagnostic process New for the Ninth Edition Features online and downloadable digital formats with rapid search functions annotation and bookmarking facilities image collections and live reference links Contains many color illustrations and high quality clinical photographs to help with interpretation and understanding Includes more than 1000 new photographs and drawings Incorporates new design elements such as alternate colour coding of chapters for easier navigation Known for its thorough yet practical approach Greenfield's continues to provide trusted information to all neuropathologists and those in related specialties including neurologists neurosurgeons general pathologists neuroradiologists and clinical neuroscientists

Neurological Therapeutics John Noseworthy,2003-05-15 A comprehensive and authoritative textbook Neurological Therapeutics Principles and Practice provides a reference that is both authoritative and accessible for daily use The textbook explores the issues underlying treatment decisions not only for the most readily treated disorders but also for those conditions with few existing definitive therapeutic options With 600 figures 37 in full color tables and a companion volume that is portable and easy to use the final product is an important reference

The Molecular and Genetic Basis of Neurologic and Psychiatric Disease Roger N. Rosenberg,2008 Completely updated for its Fourth Edition this book is the most comprehensive current review of the molecular and genetic basis of neurologic and psychiatric diseases More than 120 leading experts provide a fresh new assessment of recent molecular genetic and genomic advances offer new insights into disease pathogenesis describe the newest available therapies and explore promising areas of therapeutic development This edition features an updated section on psychiatric disease and expanded updated chapters on human genomics gene therapy

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Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease Roger N. Rosenberg, Juan M. Pascual, 2020-06-24 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease Sixth Edition Volume One provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to neurologic and psychiatric disease A favorite of over three generations of students clinicians and scholars this new edition retains and expands the informative concise and critical tone of the first edition This is an essential reference for general medical practitioners neurologists psychiatrists geneticists and related professionals and for the neuroscience and neurology research community The content covers all aspects essential to the practice of neurogenetics to inform clinical diagnosis treatment and genetic counseling Every chapter has been thoroughly revised or newly commissioned to reflect the latest scientific and medical advances by an international team of leading scientists and clinicians The contents have been expanded to include disorders for which a genetic basis has been recently identified together with abundant original illustrations that convey and clarify the key points of the text in an attractive didactic format Comprehensive coverage of the neurogenetic foundation of neurological and psychiatric disease Provides a detailed introduction on both the clinical and basic research implications of molecular and genetics surrounding the brain Includes new chapters on molecular genomics CRISPR and the most recent updates in molecular genetics

Aspartylglycosaminuria Ilkka Mononen, 1997

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