



Sphingolipids, Sphingolipidoses and Allied Disorders

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A Gutmann



Sphingolipids Sphingolipidoses And Allied Disorders:

Sphingolipids, Sphingolipidoses and Allied Disorders Bruno Volk, 2013-04-17 This text contains the scientific contributions to the Fourth International Symposium on Sphingolipids Sphingo lipidoses and Allied Disorders held at the Kingsbrook Jewish Medical Center on October 25 27 1971 These meetings were conducted under the auspices of the Isaac Albert Research Institute of the Kingsbrook Jewish Medical Center and the National Tay Sachs and Allied Diseases Association Inc Four symposia held in 1958 1961 1965 and 1971 were designed to gather the most relevant and innovative of the laboratory and field studies concerned with these hereditary disorders The texts generated by these periodic meetings have mirrored the increasing absorption of the scientific community in the problems of sphingolipid metabolism The first meeting in 1958 consisted of but twelve pre sentations the majority emanating from local laboratories The current sessions contain 48 scientific presentations by scientists from nine countries and demonstrate the increas ingly diversified techniques and approaches employed in the study of these diseases Many of the authors in exploring data on the mucopolysaccharidoses and leucodystrophies as well as the sphingolipidoses have given recognition to those biochemical areas held in common by these otherwise diverse disease processes The problems of prevention and therapy of these diseases have been considered by some of the contributors Laboratory screening procedures designed to detect carriers of the va rious lipidoses are now available and the experiences of some laboratories in this area are summarized within this volume The prospective identification of heterozygotes may indeed become a powerful adjunct in genetic counseling *Sphingolipids, Sphingolipidoses and Allied Disorders* International Symposium on the Cerebral Sphingolipidoses, 4th, Kingsbrook Jewish Medical Center, 1971, 1972 *Sphingolipids, Sphingolipidoses and Allied Disorders* Bruno Volk, 2014-01-15

Sphingolipids, Sphingolipidoses and Allied Disorders Bruno W. Volk, 1972 **Sphingolipid Biochemistry** Julian N. Kanfer, Sen-itiroh Hakomori, 2012-12-06 *The Gangliosidoses* Bruno Volk, 2012-12-06 The history of so called storage diseases goes back to the end of the 19th and to the beginning of the 20th century when Fabry Tay Sachs Gaucher Niemann Hunter and Hurler first described the disorders which up to now are called by their eponym The clinical descriptions soon were followed by pathologic studies and within a short time the hereditary characters of these rare afflictions came to be recognized Although sporadic reports during the early part of this century dealt with biochemical analysis of the stored materials in these disorders it was actually in the late 1930s that the abnormal deposits started to attract the increasing attention of chemists S H Thannhauser brought the broad concept of lipidoses as a group of related disorders to the attention of the medical profession for the first time and in 1939 Klenk observed that the brain of a patient with Tay Sachs disease contained greatly increased amounts of a glycolipid for which he proposed the name ganglioside 20 years has thrown new light on these afflic Work carried out in the past tions and has pinpointed the enzymatic and lipid abnormalities associated with the various storage diseases Moreover electron microscopic studies have permitted detailed investigations of the fine

structure of the various organs of afflicted patients National Library of Medicine Current Catalog National Library of Medicine (U.S.),1972 First multi year cumulation covers six years 1965 70 **Biology of Brain Dysfunction** Gerald E. Gaull,2012-12-06 The growth of neurochemistry molecular biology and biochemical genetics has led to a burgeoning of new information relevant to the pathogenesis of brain dysfunction This explosion of exciting new information is crying out for collation and meaningful synthesis In its totality it defies systematic summation and of course no one author can cope Thus invitations for contributions were given to various experts in areas which are under active investigation of current neurological interest and pregnant Although this project is relatively comprehensive by dint of size other topics might have been included the selection was solely my responsibility I believe systematic summation a virtual impossibility indeed hardly worth the effort The attempt to assemble all of the sections involved in a large treatise with multiple authors inevitably results in untoward delays due to the difference in the rate at which various authors work Therefore the following strategy has been adopted multiple small volumes and a relatively flexible format with publication in order of receipt and as soon as enough chapters are assembled to make publication practical and economical In this way the time lag between the ideas and their emergence in print is the shortest Scientific Directory and Annual Bibliography National Institutes of Health (U.S.),1973 *Published Scientific Papers of the National Institutes of Health* National Institutes of Health (U.S.),1973 Each issue lists papers published during the preceding year *Inborn Disorders of Sphingolipid Metabolism* Stanley M. Aronson,Bruno W. Volk,2017-01-31 *Inborn Disorders of Sphingolipid Metabolism* is a collection of papers presented at the Third International Symposium on the Cerebral Sphingolipidoses and Allied Diseases held at the Isaac Albert Research Institute of the Jewish Chronic Disease Hospital and at the State University of New York Downstate Medical Center on October 25 and 26 1965 This book is organized into three parts encompassing 35 chapters Part I deals first with electron microscopic histochemical and morphological investigations of certain sphingolipid metabolism disorders This part also examines several case reports on the features and symptoms of spongy degeneration of the central nervous system familial leukodystrophy adrenal insufficiency and cutaneous melanosis Part II surveys the metabolism biosynthesis and structure of gangliosides and sialic acids This part also considers the nature of the lipophilic portions of the brain gangliosides This part particularly looks into the features and clinical manifestation of Tay Sachs disease The third part covers the genetic and clinical aspects of the Tay Sachs disease This part also evaluates the genetics of the Hurler Hunter syndrome Batten Spielmeyer Vogt disease and lipogranulomatosis syndrome This book is of value to biochemists histochemists geneticists and researchers in the allied fields of lipidosis The Developing Brain and Its Disorders M. Arima,Y. Suzuki,H. Yabuuchi,1985-04-09 *Tay-Sachs Disease* ,2001-10-10 Tay Sachs disease is a rare hereditary disease caused by a genetic mutation that leaves the body unable to produce an enzyme necessary for fat metabolism in nerve cells producing central nervous system degeneration In infants it is characterized by progressive mental deterioration blindness paralysis epileptic

seizures and death by age four Adult onset Tay Sachs occurs in persons who have a genetic mutation that is similar but allows some production of the missing enzyme There is no treatment for Tay Sachs A test to determine whether an infant is carrying the Tay Sachs disease was introduced in 1969 However work continues to be done to help find a cure Because there is no cure for this deadly disease genetic research is essential Advances in Genetics presents an eclectic mix of articles of use to all human and molecular geneticists They are written and edited by recognized leaders in the field and make this an essential series of books for anyone in the genetics field The Glycoconjugates V2 Martin Horowitz,2012-12-02 The Glycoconjugates Mammalian Glycoproteins Glycolipids and Proteoglycans Volume II is a collaboration of different experts in the field of molecular biology on the subject of glycoconjugates The text of the second volume covers topics such as the structure and biosynthesis of connective tissue proteoglycans glycoproteins and glycolipids and glycoprotein and glycolipid catabolism and degradation It also discusses the structure composition and isolation of surface membranes and lectins its immunological aspects and its use in the study of mammalian glycoprotein The book is recommended for molecular biologists organic chemists and biochemists who would like to know more about glycolipids and glycoproteins and their applications

Lipid metabolism in mammals Fred Snyder,2012-12-06 During the past decade we have witnessed a vast expansion in our knowledge of lipid metabolism especially for mammalian tissues One obvious conclusion arising from these studies is that no single overall scheme of lipid metabolism can be classed as distinctly characteristic of all mammalian organs Although certain synthetic and degradative lipid pathways are similar in a variety of organs I have been impressed by the notable exceptions I was motivated to organize this work on Lipid Metabolism in Mammals because of the lack of a single reference source containing a comparative organ approach to lipid metabolism in mammals that emphasizes the uniqueness of pathways in the various organs of the body Because of the escalation in lipid research I also feel strongly that there is an urgent need for an updated concise account of this field The group of authors for the chapters in the two volumes of Lipid Metabolism in Mammals were selected for their expertise and personal experience with the lipid metabolism of the organs or blood constituents that are the subjects of the chapters Sufficient leeway has been given each author to approach the subject matter from a personal viewpoint However the overall direction of each chapter has been slanted to emphasize the similarities and differences in lipid metabolism among organ systems The introductory chapter on general pathways provides a convenient reference to illustrations of specific reaction sequences that are well established and that occur in a number of organs **Garner and Klintworth's Pathobiology of Ocular Disease** Gordon K. Klintworth,Alec Garner,2008-03-27

Moving from a standard clinical reference Garner and Klintworth's Third Edition of Pathobiology of Ocular Disease is now modeled to be both a scientific research tool and a quick reference option for the practicing ophthalmologist ophthalmic pathologist optometrist and vision scientist With the addition of ten new chapters this edition focuses **Genetic**

Determinants of Pulmonary Disease Stephen. D. Litwin,2020-08-13 This book provides a current and integrated approach

to the subject of genetic determinants of pulmonary disease with emphasis on physiologic derangements and genetic mechanisms It describes the epidemiologic genetic approach to chronic pulmonary disease

The Neuronal Ceroid Lipofuscinoses (Batten Disease) Hans H. Goebel, Sara E. Mole, Brian D. Lake, 1999 The European Concerted Action on the Neuronal Ceroid Lipofuscinoses NCL probably the most frequent group of progressive neurodegenerative disorders in children has been the subject of a 3 year European Concerted Action ECA NCL within the Biomed 2 programme listed among Rare diseases The NCL are a lysosomal disease which have recently been confirmed by important genetic studies on the NCL which revealed a diversity of at least 8 gene loci related to this group of diseases ECA NCL represents a multi national multi disciplinary network of researchers on the NCL which have provided new data on this group of diseases In this book clinical features neuroradiological and electrophysiological data morphological aspects genetic data and biochemical results will be provided by actively engaged researchers on NCL from this Concerted Action who have also provided major presentations at the two recent International Congresses of NCL 1996 in Helsinki NCL 96 and 1998 in Dallas NCL 98 Thus this book represents a state of the art written both for active researchers on NCL and other lysosomal and neurodegenerative diseases as well as for those interested in NCL for other reasons foremost patients and their parents Tables illustrations and most recent references will accomplish the text produced and edited by an editorial board consisting of participants of this ECA NCL At the end of this state of the art presentation future lines of research including therapies will be outlined and emphasized what will have to be done to arrive at complete elucidation of this group of progressive neurodegenerative disorders

The Cultured Cell and Inherited Metabolic Disease R. Angus Harkness, F. Cockburn, 2012-12-06 The use of cultured cells in the clinical diagnosis of hereditary metabolic disease is a rapidly developing subject to which many different disciplines have brought their expertise and knowledge A number of scientists who have individually contributed to the growth of the subject gave invited papers at the Fourteenth Symposium of the Society for the Study of Inborn Errors of Metabolism in the University of Edinburgh on 13-16th July 1976 These papers form the basis of this monograph which brings together contributions from the basic sciences and from physicians concerned primarily with human disease The cross fertilization produced by this interdisciplinary communication was invaluable to those trying to understand and overcome diagnostic problems posed by hereditary metabolic disease Cell culture methods and cell preservation techniques were described by D G Harnden and D E Pegg Dr T Elsdale outlined some of the factors which control in vitro cell growth and division Cell culture methods and cryopreservation techniques have allowed the wide distribution of biochemically abnormal cells and their study over long periods of time It is also evident that when a defect which produces severe metabolic disorder in man can be studied in the laboratory using isolated cell cultures a wide variety of investigative procedures can be focused on to the cellular defect without distress or discomfort to the patient or relatives

Tumors of the Bone Marrow Richard D. Brunning, Robert W. McKenna, 1994

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