

Mitochondrial Complex I Deficiency

Kenneth F. Swaiman, Stephen Ashwal, Donna M Ferriero, Nina F Schor, Richard S. Finkel, Andrea L. Gropman, Phillip L. Pearl

Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease Roger N. Rosenberg, Juan M. Pascual, 2014-10-28 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease, Fifth Edition provides a comprehensive introduction and reference to the foundations and key practical aspects relevant to the majority of 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. Previous editions have established this book as the leading tutorial reference on neurogenetics. Researchers will find great value in the coverage of genomics, animal models and diagnostic methods along with a better understanding of the clinical implications. Clinicians will rely on the coverage of the basic science of neurogenetics and the methods for evaluating patients with biochemical abnormalities or gene mutations, including links to genetic testing for specific diseases. - Comprehensive coverage of the neurogenetic foundation of neurological and psychiatric disease - Detailed introduction to both clinical and basic research implications of molecular and genetic understanding of the brain - Detailed coverage of genomics, animal models and diagnostic methods with new coverage of evaluating patients with biochemical abnormalities or gene mutations

Mitochondrial Complex I Deficiency Elena Jane Tucker, 2011

Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases Nenad Blau, Carlo Dionisi Vici, Carlos R. Ferreira, Christine Vianey-Saban, Clara D. M. van Karnebeek, 2022-02-21 This updated and enlarged second edition is a unique source of information on the diagnosis, treatment, and follow-up of metabolic diseases. The clinical and

laboratory data characteristic of rare metabolic conditions can be bewildering for clinicians and laboratory personnel alike – reference laboratory data is scattered, and clinical descriptions can be obscure. The new Physician's Guide with the additional more than 600 diseases now featured, documents 1200 conditions grouped according to type of disorder, organ system affected (e.g. liver, kidney, etc) or phenotype (e.g. neurological, hepatic, etc). It includes relevant clinical findings and highlights the pathological values for diagnostic metabolites. Guidance on appropriate biochemical genetic testing is also provided and established experimental therapeutic protocols are described, with recommendations on follow-up and monitoring. The authors are acknowledged experts, and the book is a valuable desk reference for all who deal with inherited metabolic diseases. Chapter 73 is available open access under a Creative Commons Attribution 4.0 International License via link.springer.com

Magnetic Resonance of Myelination and Myelin Disorders Marjo S. van der Knaap, Jaap Valk, 2005-08-15 Our thanks go to our colleagues at the VU University Medical Center and to those in other hospitals Reading through the prefaces of the two previous editions, we can say that much of what was said there still holds. At the same time, however, much has changed. published or unpublished, making it possible for us to There has been immense progress in the technical present illustrations of nearly all known white matter possibilities of magnetic resonance and in the know-disorders. Two colleagues were particularly helpful ledge of genetic defects, biochemical abnormalities, and provided us with essential and unpublished f- and cellular processes underlying myelin disorders. ures: our friends Susan Blaser, from the Hospital for This immense progress has prompted us to embark Sick Children in Toronto, and Zoltán Patay, from the upon the enormous task of rewriting the previous King Faisal Hospital in Riyadh. edition and adding 40 chapters. In doing so we have Many people at the VU University Medical Center tried to cover most white matter disorders, hereditary have been of great technical help to us in producing and acquired, and to present a collection of images to high quality images and in providing secretarial illustrate the field to the fullest possible extent. This assistance. The contributions of these people are edition will therefore be more complete than the pre- mentioned separately in the acknowledgements.

Mitochondrial Complex I Deficiency Federica Valsecchi, 2012

Principles and Practice of Clinical Cardiovascular Genetics Dhavendra Kumar, Perry Elliott, 2010-04-02

Mitochondrial Complex I Deficiency Results in Neuronal Dysfunction, Degeneration, and Death Shane E. Kruse, 2009

Swaiman's Pediatric Neurology E-Book Kenneth F. Swaiman, Stephen Ashwal, Donna M Ferriero, Nina F Schor, Richard S. Finkel, Andrea L. Gropman, Phillip L. Pearl, 2017-03-16 Since 1975, Dr. Kenneth Swaiman's classic text has been the reference of choice for authoritative guidance in pediatric neurology, and the 6th Edition continues this tradition of excellence with thorough revisions that bring you fully up to date with all that's new in the field. Five new sections, 62 new chapters, 4 new

editors, and a reconfigured format make this a comprehensive and clearly-written resource for the experienced clinician as well as the physician-in-training. - Nearly 3,000 line drawings, photographs, tables, and boxes highlight the text, clarify key concepts, and make it easy to find information quickly.

The Molecular and Genetic Basis of Neurological Disease Roger N. Rosenberg,1993

Developmental Disabilities in Infancy and Childhood: Neurodevelopmental diagnosis and treatment Arnold J. Capute,Pasquale J. Accardo,1996 The second edition, now in two volumes rather than one, contains some 30 new chapters addressing a greatly increased number of topics and range of issues, from a variety of disciplines. The first volume offers discussion of general issues in the theory, assessment, and treatment of developmental dis

Cell Biological Consequences of Inherited Mitochondrial Complex I Deficiency Sjoerd Adrianus Johannes Verkaart,2007

The Scientific Basis for the Treatment of Parkinson's Disease Charles Warren Olanow,Abraham N. Lieberman,1992 An international review of recent methods of treating Parkinson's disease, and the scientific basis on which these methods are based. At the same time, the book covers preventative and grafting treatment.

Syndromes: Rapid Recognition and Perioperative Implications, 2nd edition Bruno Bissonnette,2019-09-13 Publisher's Note: Products purchased from Third Party sellers are not guaranteed by the publisher for quality, authenticity, or access to any online entitlements included with the product.The ULTIMATE reference about syndromes to have on hand to facilitate rapid recognition and perioperative implications --- in full colorSyndromes: Rapid Recognition and Perioperative Implications, Second Edition, is the ultimate reference for anesthesiologists, surgeons, internists, pediatricians, emergency physicians and nurses caring for patients affected with special operative needs and acute medical care. This updated book provides a complete A-to-Z encyclopedic clinical presentation of more than 2,200 syndromes. Extensive cross-referencing of international synonyms, classifications, and other conditions to be considered in the differential diagnosis further expands coverage to more than 5,000 entries. It is the most comprehensive reference on the subject. Syndromes: Rapid Recognition and Perioperative Implications, Second Edition, is enhanced by 200 high-quality photographs, in addition to tables, classifications and clinical management considerations. The addition of color tabs, a new feature, improve consultation and speed information retrieval. Each syndrome, listed in alphabetical order, is described as follows:•“At a glance” description•Synonyms•Incidence•Genetic Inheritance•Pathophysiology•Diagnosis•Clinical Aspects•Precautions Before Anesthesia•Anesthetic Considerations•Pharmacological Implications•Other conditions to be considered•ReferencesHealth care providers, fellows and residents in training will find this textbook to be an essential reference to improve patient care in this highly demanding medical and surgical environment.

Mitochondrial Disorders in Neurology Anthony Henry Vernon Schapira,S. DiMauro,1994 Mitochondrial Disorders in Neurology provides an overview of mitochondrial diseases. This book discusses the effects of mitochondrial dysfunction

based on the relevant biochemistry and molecular genetics. The abnormal muscle and mitochondrial morphology in a variety of clinical presentations from isolated ophthalmoplegia to severe encephalopathy are also elaborated. This text likewise deliberates Leber's hereditary optic neuropathy, neurodegenerative disorders, and respiratory chain defects. Other topics covered include mitochondrial DNA and the genetics of mitochondrial disease; cytochrome ox ...

Muscular Dystrophy Abstracts ,1989

Mitochondrial Disorders Caused by Nuclear Genes Lee-Jun C. Wong,2012-09-18 Mitochondrial cytopathies are mutations in the inherited maternal mitochondrial genome, or the nuclear DNA-mutation. Mitochondrial respiratory chain disorders (RCD) are a group of genetically and clinically heterogeneous diseases, due to the fact that protein components of the respiratory chain are encoded by both mitochondrial and nuclear genomes and are essential in all cells. In addition, the biogenesis, structure and function of mitochondria, including DNA replication, transcription, and translation, all require nuclear encoded genes. Since mitochondria are present in every cell, every tissue, mitochondrial disorder usually affects multiple organs.

Biological Signals and Receptors ,2001

Hurst's the Heart, 14th Edition: Two Volume Set Valentin Fuster,Robert A. Harrington,Jagat Narula,Zubin J.

Eapen,2017-03-22 Cardiology's cornerstone text - thoroughly updated to reflect the latest clinical perspectives and cutting-edge topics Hailed for its authority, currency, and ability to translate the latest technical and clinical advances into clinical application, Hurst's The Heart is the field's landmark text and cardiology's longest continuously published reference text. A previous edition of this trusted classic was described by Doody's as "an outstanding choice for those who strive for a firm foundation in cardiovascular medicine, as well as an up-to-date and user-friendly source that addresses every discipline in the field." Readers will find succinct, visually appealing summaries of all the major new trials, and guidelines, along with tips for optimizing outcomes and health quality. The Fourteenth Edition has been completely updated to reflect the latest technical, therapeutic, and clinical advances, while still maintaining a strong focus on patient care. Other enhancements include the addition of textual features such as Practice Points, Common Clinical Questions, and an increased number of the acclaimed Hurst's diagrams. • NEW TO THIS EDITION: Section on Metabolic Disorders and Cardiovascular Disease, Cigarette Smoking and Cardiovascular Disease • Enhanced by more than 1,500 full-color illustrations and more than 500 tables • Brand new chapters include: Arrhythmogenic Cardiomyopathy, Ischemic Mitral Regurgitation, Degenerative Mitral Valve Disease, Left Ventricular Noncompaction, Evaluation and Management of Acute Heart Failure, Carotid Artery Stenting, Race, Ethnicity, and Cardiovascular disease • The only comprehensive cardiology reference to publish every 2.5 years to allow for reporting of the latest trials and guidelines

Neuropediatrics ,1988

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