

Mitochondrial Complex I Deficiency

Marjo S. van der Knaap,Jaap Valk

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

Rosenberg's Molecular and Genetic Basis of Neurological and Psychiatric Disease, Seventh Edition Roger N. Rosenberg,Juan M. Pascual,2024-10-07 Rosenberg's Molecular and Genetic Basis of Neurologic and Psychiatric Disease, Seventh Edition provides a comprehensive introduction and reference to the foundations and practical aspects relevant to the majority of neurologic and psychiatric disease. This updated volume focuses on degenerative disorders, movement disorders, neuro-oncology, neurocutaneous disorders, epilepsy, white matter diseases, neuropathies and neuronopathies, muscle and neuromuscular junction disorders, stroke, psychiatric disease, and a neurologic gene map. This volume includes new

chapters on Von Hippel-Lindau disease, antisocial and violent behavior and Autism. A favorite of over four generations of students, clinicians and scholars, this new edition retains and expands on the informative, concise and critical tone of the previous edition. This is an essential reference for general medical practitioners, neurologists, psychiatrists, geneticists, related professionals, and for the neuroscience and neurology research community at large. - Both volumes combined provide a comprehensive coverage on the neurogenetic foundation of neurological and psychiatric disease - This volume presents detailed coverage of disease mechanisms, and management for degenerative disorders, movement disorders, and muscle and neuromuscular junction disorders. - Includes new chapters on the pharmacogenomics of Alzheimer's Disease and Epilepsy and the most recent updates in molecular genetics, focusing on pain genetics and muscular dystrophy

Inborn Errors of Metabolism Brendan H. Lee, Fernando Scaglia, 2015 Texts on inborn errors of metabolism (IEMs) have traditionally focused on classical biochemistry, clinical presentation, and standard treatment approaches. Inborn Errors of Metabolism is an expansion on this model, one that establishes an innovative pathway approach and provides a new authority on this family of disease. Alongside the standard cadre of molecular and clinical underpinnings, this volume includes coverage of newborn screenings and an overarching treatment of IEMs as complex diseases - how basic alterations can lead to complex secondary and tertiary effects in metabolism that contribute to increasingly recognized natural histories of disease.

Mitochondrial Complex I Deficiency Elena Jane Tucker, 2011

The Dynamic Nature of Mitochondria Andreas S. Reichert, 2024-04-15 Mitochondrial research has exploded over the last ~150 years. This book gives an amazing view on a conceptual change in our understanding of mitochondrial biology. It becomes clear that mitochondria are extremely dynamic in nature, controlling life at multiple levels. Mitochondria rule energy conversion, adapt cells well to changing stress and nutrient conditions, and regulate many cellular processes including immunity. The dynamic nature of mitochondria occurs at an intramitochondrial level but also includes its ability to interact with other organelles and to modulate multiple signalling pathways. It is thus not surprising that alterations or inabilities to ensure this dynamic behaviour is linked to ageing and human diseases. The following sections give an updated view on mitochondria: Mitochondrial ultrastructure: molecular mechanisms shaping the inner membrane Mitochondrial cristae and lipid dynamics: from super-resolution microscopy to lipid-OXPHOS interplay Mitochondrial control of cellular homeostasis: from redox signalling to interorganellar contact sites Mitochondria in health and disease: from mtDNA release to Complex I assembly Advanced methods in mitochondrial biology and metabolism research Integrative view on mitochondrial research and outlook The field of mitochondrial research has always been full of surprises and has helped science to advance tremendously. It developed hand in hand with landmark developments in technology, such as super-resolution microscopy (nanoscopy), and is currently influencing an increasing number of scientific disciplines. There is still

much 'new' to find out about this 'old' organelle and I think that you can find interesting and also unexpected aspects of mitochondrial biology in this book. I hope the book will enhance your scientific curiosity and inspire your own research.

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

Mitochondrial Complex I Deficiency Federica Valsecchi, 2012

Inborn Metabolic Diseases John Fernandes, Jean-Marie Saudubray, Georges van den Berghe, John H. Walter, 2006-11-22 This classical textbook has become indispensable for those in the front line dealing with metabolic disorders. The book is aimed at all those involved with this specialty including pediatricians, biochemists, dieticians, neurologists, internists, geneticists, psychologists, nurses, and social workers. This 4th edition has been thoroughly updated and revised. One new chapter on Neonatal screening by tandem MS/MS has been added and several new groups of disorders have been included. The book's main feature is the strong emphasis on clinical presentation and treatment in acute and chronic situation.

Myopathology Balan Louis Gaspar, Rakesh Kumar Vasishta, Bishan Dass Radotra, 2018-09-24 This book covers all aspects of basic, essential, recent advances and controversies in myopathology. The major emphasis is on diagnostic myopathology of muscular dystrophies, inflammatory myopathies, mitochondrial myopathies, metabolic myopathies, congenital myopathies, myopathies of miscellaneous etiology, neurogenic and neuromuscular junction disorders, the goal being to broaden readers' understanding of individual disease subgroups. The book also contains all the essential details needed to establish a neuromuscular lab, making it especially relevant for laboratory technical staff and research scholars.

Neurology of the Newborn Joseph J. Volpe, 2008-01-01 Provides the insights in neonatal neurology. This title describes from the discoveries in genetics through the advances in the diagnosis and management of neurologic disorders. It delivers clinical guidance you need to provide effective care for neonates with neurological conditions.

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 who referred their patients to us. We are indebted to them, 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 knowledge of genetic defects, biochemical abnormalities, and provided us with essential and unpublished data and cellular processes underlying myelin disorders. We are indebted to our friends Susan Blaser, from the Hospital for Sick Children in Toronto, and Zoltán Patay, from the King Faisal Hospital in Riyadh, upon the enormous task of rewriting the previous 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 acquiring, and to present a collection of high quality images and in providing secretarial assistance to illustrate the field to the fullest possible extent. The contributions of these people are therefore more complete than the pre-mentioned separately in the acknowledgements.

Human Drug Targets Edward D. Zanders, 2015-11-09 The identification of drug targets in a given disease has been central to pharmaceutical research from the latter half of the 20th century right up to the modern genomics era. Human Drug Targets provides an essential guide to one of the most important aspects of drug discovery – the identification of suitable protein and RNA targets prior to the creation of drug development candidates. The first part of the book consists of introductory chapters that provide the background to drug target discovery and highlight the way in which these targets have been organised into online databases. It also includes a user's guide to the list of entries that forms the bulk of the book. Since this is not designed to be a compendium of drugs, the emphasis will be on the known (or speculated) biological role of the targets and not on the issues associated with pharmaceutical development. The objective is to provide just enough information to be informative and prompt further searches, while keeping the amount of text for each of the many entries to a minimum. Human Drug Targets will prove invaluable to those drug discovery professionals, in both industry and academia, who need to make some sense of the bewildering array of online information sources on current and potential human drug targets. As well as creating order out of a complex target landscape, the book will act as an ideas generator for potentially novel targets that might form the basis of future discovery projects.

Clinical Studies and Therapies in Parkinson's Disease Juan Segura-Aguilar, 2021-06-12 More than 50 years have passed since the use of L-dopa in the palliative treatment of Parkinson's disease, but it remains the most common treatment despite inducing severe side effects such as dyskinesia after 4–6 years of use. Numerous preclinical investigations based on

endogenous neurotoxin models have promised various therapies for Parkinson's disease, but these efforts have failed when attempting to transfer these successful results to preclinical studies. Although several publications have warned of these failures, the scientific community remains mostly unaware, and there is a need to focus their efforts on potential therapeutics that can slow or halt development of the disease. *Clinical Studies and Therapies in Parkinson's Disease: Translations from Preclinical Models* analyzes preclinical models based on exogenous neurotoxins and why they have failed. Neuroscientists, neurologists, and neuropharmacologists will benefit greatly from the book's discussion of these newer models, their benefits, and the need for their implementation. This book also provides the basic concepts of dopamine metabolism for students taking courses in neurochemistry, neuroscience, neuropharmacology, biochemistry, and medicine. - Reviews Parkinson's disease classification, pharmacological therapies, and nonmotor and motor symptoms - Analyzes preclinical models of Parkinson's disease therapies based on exogenous neurotoxins and why they have failed - Reviews genetic preclinical models based on genetic mutations and endogenous neurotoxins - Proposes a more physiological model directly related to the metabolism of dopaminergic neurons - Provides the basic concepts and mechanisms of dopamine metabolism

JIMD Reports - Volume 12 Johannes Zschocke, K Michael Gibson, Garry Brown, Eva Morava, Verena Peters, 2014-07-08
JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder, or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.

Inherited Neuromuscular Diseases Carmen Espinós, Vicente Felipo, Francesc Palau, 2010-03-11 This reference on the state-of-the-art of neuromuscular diseases as a whole offers a current review of inherited neuromuscular diseases under different approaches: genetics, pathomechanisms, therapies and treatments.

Swaiman's Pediatric Neurology - E-Book Stephen Ashwal, Phillip L. Pearl, 2025-05-09 **Selected for 2025 Doody's Core Titles® in Pediatrics and with Essential Purchase designation in Neurology** For fifty years, experienced clinicians and physicians in training have relied on Swaiman's cornerstone text as their #1 source for authoritative guidance in pediatric neurology. *Swaiman's Pediatric Neurology: Principles and Practice, Seventh Edition*, continues this tradition of excellence under the expert editorial direction of Drs. Stephen Ashwal and Phillip L. Pearl, along with a team of key leaders in the field who serve as associate and section editors in their areas of expertise. Thorough revisions—including new chapters, new videos, new editors, and expanded content—bring you up to date with this dynamic field. - Contains new sections on global child neurology and environment and brain development and a greatly expanded section on neurogenetics, in addition to new chapters on autoimmune epilepsies, immune-mediated movement disorders, and more. - Offers expanded online content, including additional figures, tables, and text, as well as new personal introductory videos by many chapter authors. - Covers new, emerging, or controversial topics such as COVID-19, teleneurology, environment and brain development, immune-

mediated disorders of the nervous system, functional neurological disorders in children, nonverbal learning disorders, and the pharmacological and future genetic treatment of neurodevelopmental disabilities. - Provides authoritative coverage of perinatal acquired and congenital disorders, neurodevelopmental disabilities, extensive sections on pediatric epilepsy and movement disorders, nonepileptiform paroxysmal disorders, and disorders of sleep. - Features nearly 3,000 line drawings, photographs, tables, and boxes that highlight the text, clarify key concepts, and make it easy to find information quickly.

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JIMD Reports, Volume 15 Johannes Zschocke, K. Michael Gibson, Garry Brown, Eva Morava, Verena Peters, 2014-12-18 JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.

Parkinson's Disease Stewart Factor, William J. Weiner, 2007-12-15 Parkinson's disease (PD) is the second most common neurodegenerative disease in the world. Still the only major text on the subject, the completely revised and updated second edition of *Parkinson's Disease: Diagnosis and Clinical Management* comes at a time when specialists have made important advances in our understanding of the etiology, pathogenesis, investigation, and management of Parkinson's disease. The book includes 23 completely new chapters, and has updated information on: Genetics Pathology Biomarkers Pathogenesis Impulse control disorders in Parkinson's disease Updated outcome measures Complementary and alternative medicine for the treatment of Parkinson's disease Together the chapters form a comprehensive review of the many issues facing PD physicians today. Lucid and easily readable from beginning to end, each chapter may also stand on its own as a scholarly review of the individual subject. Each one is concisely written and heavily referenced for this purpose. The second edition of *Parkinson's Disease: Diagnosis and Clinical Management* provides a state-of-the-art review of where we've been, where we are now, and where we are going in treating this disease.

Motor System Disorders, Part I David S. Younger, 2023-08-08 *Motor System Disorders: Normal Physiology and*

Function and Neuromuscular Disorders summarizes recent advances and best practice for understanding normal physiology and function of the larger motor system as well as the diagnosis, course, and treatment of neuromuscular disorders. The work will include neuroanatomy, circuits and synapses. It discusses how different parts of the brain contribute to motor control, including both vestibular and cerebellar motor control, as well as the influences of the parietal and frontal lobes. Diagnostic methods include genetic evaluation, electrodiagnosis, pathology, and brain imaging. Neuromuscular disorders of both children and adults will be included. - Identifies normal motor control physiology and function, including neuroanatomy, circuits, and synapses - Discusses vestibular and cerebral motor control and contributions of parietal and frontal lobes - Provides diagnostic methods, including genetic evaluation, electrodiagnosis, brain imaging, and pathology analyses - Summarizes the diagnosis, course, and best practice treatment of neuromuscular disorders - Explores neuromuscular disorders in adults and children

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