

SECOND EDITION

Neuromuscular Disease

A Case-Based Approach



Jessica E. Hoogendijk, Marianne de Visser,
Pieter A. van Doorn, and Erik H. Niks

CAMBRIDGE

Medicine

Neuromuscular Disease A Case Based Approach

Reggie C. Hamdy, Neil Saran



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Neuromuscular Disease Jessica E. Hoogendijk, Marianne de Visser, Pieter A. van Doorn, Erik H. Niks, 2024-12-19 There are over 800 neuromuscular disorders and this can leave clinicians feeling lost as they try to diagnose and manage patients On the basis of 66 adult and paediatric neuromuscular case vignettes readers will be walked through using case histories and clinical manifestations as a starting point for diagnosis For each case diagnostic tools disease pathogenesis prognosis and treatment options are discussed Symptoms signs and syndromes are cross linked to help the reader navigate the array of disorders Accompanying tables explain differential diagnoses and 30 videos demonstrate clinical features This second edition has been thoroughly updated as the neuromuscular subspecialty has developed from clinically and pathologically descriptive to making use of new diagnostic technologies and therapies Neurologists at all levels paediatricians internists geneticists rehabilitation physicians physiotherapists and researchers in the field will find this an invaluable guide as they seek to familiarise themselves with this complex range of disorders

Neuromuscular Disease John H. J. Wokke, Pieter A. van Doorn, Jessica E. Hoogendijk, Marianne de Visser, 2013-03-07 There are over 600 neuromuscular disorders and the variability of these syndromes can leave clinicians feeling as if they are lost in a maze as they seek to diagnose and manage patients This book addresses this problem by using the case history and symptom manifestation as a starting point for the diagnostic process in adult patients mimicking the situation in the consultation room For each case diagnostic tools disease pathogenesis prognosis and treatment options are discussed along with rare manifestations and differential diagnoses Symptoms signs and syndromes are cross linked to help the reader navigate the variety of disorders Accompanying tables give a broader picture of the manifestations of a particular disease within the landscape of neuromuscular disorders This highly illustrated book with accompanying videos will aid neurologists at all levels internists geneticists rehabilitation physicians and researchers in the field as they seek to familiarize themselves with this complex range of disorders
Genetic Neuromuscular Disorders Corrado Angelini, 2017-10-05 This updated and expanded new edition of a successful book describes genetic diagnostic entities of neuromuscular disorders Neuromuscular syndromes are presented clinically either as a case study or as an overview from the literature accompanied by text presenting molecular defects and differential diagnosis This collection of neuromuscular disorders features the differential clinical phenotypes related to each genotype and are representative of the whole spectrum of a genetic muscle disorder helping the clinician and neuromuscular physician to make a diagnosis Key points for each genetic disease are identified to suggest treatment when available or the main clinical exams useful in follow up of patients *Genetic Neuromuscular Disorders A Case Based Approach* is aimed at neuromuscular physicians and neurology residents Interpretation of Complex

Arrhythmias: A Case-Based Approach, An Issue of Cardiac Electrophysiology Clinics Melvin Scheinman, 2016-03-12 This special case based issue of Cardiac Electrophysiology Clinics features a selection of cases on the following topics Use of the Surface ECG to Define the Nature of Challenging Arrhythmias Diagnostic and Prognostic Implications of Surface Recordings from Patients with A V Block Supraventricular Tachycardia Ventricular Tachycardia Atrial Fibrillation and Flutter Troubleshooting Device Function Adult Congenital Cardiac Disease and Arrhythmias in Patients with Genetic Arrhythmia Syndromes

Essentials Of Orthopaedics For Undergraduates: A Case-based Approach Joseph Thambiah, Eng Hin Lee, James Hui Hoi Po, 2025-04-08 A concise textbook of orthopaedics that uses real life situations and clinical presentations to teach analytical skills in the diagnosis and management of common orthopaedic conditions contextualised for an Asian practice The book covers commonly encountered conditions and teaches the skills to reach a reasoned differential diagnosis and craft an appropriate management plan It also discusses consistent physical examination skills including with video demonstrations by eminent faculty As in the book description above contributors are from across Singapore The book deals with commonly encountered clinical orthopaedic situations and diagnoses rather than esoteric and rare conditions It is therefore a valuable resource for junior doctors and family practitioners as well

A Case-Based Guide to Neuromuscular Pathology Lan Zhou, Dennis K. Burns, Chunyu Cai, 2019-10-24 This book provides a practical guide to the field of neuromuscular pathology including muscle peripheral nerve and skin biopsies with biopsy interpretation Divided into three parts the book begins with introductory chapters that review muscle nerve and skin biopsy indications and procedures normal muscle and nerve histology and common muscle and nerve pathology The 28 myopathy case chapters in Part 2 and 11 neuropathy case chapters in Part 3 illustrate the clinical and pathological features of many neuromuscular diseases demonstrate the indications and utilities of biopsies discuss clinical and pathological differential diagnosis update the individual disease management and summarize pertinent clinical and pathology pearls for each case A Case Based Guide to Neuromuscular Pathology will function as an indispensable reference for neurologists and pathologists looking to understand the utility of muscle nerve and skin biopsies This book will also aid neurology residents neuromuscular fellows and neuropathology fellows in preparing for questions related to the muscle and nerve pathology in their in service and board exams

Case-based Approach to Common Neurological Disorders Krishna Kumar Oli, Gentle Sunder Shrestha, Rajeev Ojha, Pramod Kumar Pal, Sanjay Pandey, Bibhukalyani Das, 2024-03-13 This book primarily aims to assist doctors from non neuroscience background to acquire knowledge about common neurological disorders encountered in a resource constrained setup in a case based and practical approach The book elucidates the value of proper and systematic approach to the patient how proper history taking and clinical evaluation followed by appropriate investigations can narrow down the differential diagnoses and help proper and timely management of the patients The book covers most of the common neurological disorders and scenarios encountered in clinical practice and will benefit clinicians facing difficult cases particularly those

involving fundamental research and diagnostic methods This book is useful for physicians working in internal medicine physicians handling neurology cases in resource constrained facilities and fellows in neurology The contents of this book support and practically enhance the knowledge acquired from other text books *Questions And Answers In*

Neuro-ophthalmology: A Case-based Approach Andrew G Lee, Nagham Al Zubidi, Arielle Spitze, Sushma

Yalamanchili, 2014-07-11 The book presents the most common problems seen in neuro ophthalmology in a case based learning format that provides rapid access and is easy to read The user friendly format guides and stimulates the reader to identify the distinctive symptoms in the main complaint and history to define the differentiating signs on neuro ophthalmic exam and to describe the key laboratory or neuroimaging in specific entities It also challenges the reader to ask additional questions of themselves and others by providing an initial set of questions and answers on specific neuro ophthalmologic topics The questions chosen are meant to simulate real world questions in the clinic the answers given serve to provide both the rationale for the correct answer and the explanations or reasons for the incorrect but plausible answers that might distract or mislead a clinician in specific neuro ophthalmic cases *Pediatric Kidney Disease: A Case-Based Approach -*

E-Book Farahnak Assadi, 2024-07-24 An ideal resource for specialists and non specialist clinicians and trainees *Pediatric Kidney Disease A Case Based Approach* contains more than 400 case reports that offer a unique learning opportunity in this challenging field These accessible and instructive case studies help readers recognize important clinical symptoms and signs to develop the diagnostic and management skills needed for treating patients in a routine clinical practice With wide ranging coverage of the kidney related complaints most likely to be encountered this volume is an excellent reference or certification review tool for residents fellows and clinicians who need quick access to current scientific and clinical information on managing pediatric kidney disease Provides complete state of the art reviews and assessments of kidney disease in children in a concise templated format Presents more than 400 cases covering acute and chronic kidney failures congenital and hereditary kidney diseases glomerular and vascular diseases hereditary kidney disorders hypertension tubulointerstitial diseases and urinary tract infections Begins each case with a succinct summary of patient's history signs and symptoms examinations and initial investigations followed by questions and answers Provides answers and comments that offer a detailed discussion on each topic with further case references where appropriate An eBook version is included with purchase The eBook allows you to access all of the text figures and references with the ability to search customize your content make notes and highlights and have content read aloud Additional digital ancillary content may publish up to 6 weeks following the publication date [Peripheral Nerve and Muscle Disease](#) Jeffrey A. Cohen, Jeffrey Allen Cohen, Justin Mowchun, Jon

Grudem, 2009-06-03 This book is a case based learning guide for neuromuscular diseases This second volume in the What Do I Do Now series *Peripheral Nerve and Muscle Disease* recreates real clinical situations in which the attending is asked for advice as the neurological consultant The approach to the clinical question is discussed emphasizing differential diagnosis

and treatment pathways The authors have included clinical pitfalls which they have experienced in their own practice This is a readable book which can be picked up in spare moments Palliative Medicine: A Case-Based Manual Susan

MacDonald, Leonie Herx, Anne Boyle, 2021-12-08 Palliative medicine is the medical care provided for people who have a life limiting illness or condition It encompasses both the physical symptoms a person may experience but also the psychosocial emotional and ethical issues that may arise Now in its fourth edition Palliative Medicine A Case Based Manual walks clinicians through the management of the most common situations found in palliative medicine from diagnosis and managing symptoms through to grief and bereavement Using real patient case scenarios and an easy to read question and answer format it gives students and medical professionals an accessible evidence based entryway to gain the skills and knowledge needed to provide high quality palliative and end of life care to patients and their families This new edition has been fully updated to cover the latest guidance including new chapters dedicated to medical cannabis opioids grief and bereavement and wider issues including palliative care in the community structurally vulnerable populations and public health Written by over 50 experienced palliative care clinicians and educators this book will be a welcome guide for palliative care workers medical and nursing students and medical professionals of any specialty where palliative needs might arise *A Case-Based*

Approach to Knee Pain Michelle Leong, Grant Cooper, Joseph E. Herrera, Peter Murphy, 2022-12-25 Knee pain is one of the most common reasons for patient visits to orthopedic physiatrist primary care and sports medicine offices Most books that cover this topic review it as a chapter within a larger book on orthopedics as a whole or they focus on one specific aspect of knee pathology such as osteoarthritis This practical text is an evidence based user friendly review of the literature for the breadth of knee pathologies that present to the busy practitioner Opening with a review of the relevant anatomy subsequent chapters discuss injuries to ligaments and tendons the meniscus and osteoarthritis Additional chapters cover knee pain in the pediatric patient sports trauma and fractures and rheumatologic and infectious disease considerations And while reviewing pathology and its diagnosis and treatment is important proceeding through real case studies is extremely valuable in bringing the diagnosis and treatment of knee pathologies to life hence an engaging section of clinical case material rounds out the presentation Taken together A Case Based Approach to Knee Pain will be an ideal resource for musculoskeletal medicine practitioners of all types

Ruppel's Manual of Pulmonary Function Testing - E-Book Carl Mottram, 2017-01-11 NEW UPDATED content reflects the latest guidelines testing procedure recommendations and interpretive strategies of the American Thoracic Society European Respiratory Society as well as the newest guidelines for exercise testing from the American Thoracic Society American College of Chest Physicians NEW Practice tests on the Evolve companion website help you apply the knowledge learned in the text NEW Summary Points at the end of chapters reinforce important entry level and advanced level concepts *Neuromuscular Disease with Online Resource* Jessica E.

Hoogendijk, Marianne de Visser, Pieter A. van Doorn, Erik H. Niks, 2025-02-28 There are over 800 neuromuscular disorders

and this can leave clinicians feeling lost as they try to diagnose and manage patients On the basis of 66 adult and paediatric neuromuscular case vignettes readers will be walked through using case histories and clinical manifestations as a starting point for diagnosis For each case diagnostic tools disease pathogenesis prognosis and treatment options are discussed Symptoms signs and syndromes are cross linked to help the reader navigate the array of disorders Accompanying tables explain differential diagnoses and 30 videos demonstrate clinical features This second edition has been thoroughly updated as the neuromuscular subspecialty has developed from clinically and pathologically descriptive to making use of new diagnostic technologies and therapies Neurologists at all levels paediatricians internists geneticists rehabilitation physicians physiotherapists and researchers in the field will find this an invaluable guide as they seek to familiarise themselves with this complex range of disorders

Practical Management of Pediatric Ocular Disorders and Strabismus Elias Traboulsi, Virginia Utz, 2016-07-27 This case based approach to the management of pediatric eye diseases and strabismus teaches the novice as well as the experienced surgeon how to conceptualize and analyze surgical planning to maximize patient outcomes allowing readers to get inside the head of the surgeon Individual cases with clear instructions and an abundance of color illustrations presented by experts in the field teach critical information one case at a time Practical Management of Pediatric Ocular Disorders and Strabismus provides the key guiding principles of a traditional textbook in an easy to digest format Each chapter of the text tackles a common or rare clinical situation and begins with the description of an illustrative case The chapters are structured with sections on case description differential diagnosis management and outcomes with a list of possible complications and the approach to their management

Practical Approach to Pediatric Intensive Care Praveen Khilnani, 2023-10-31 This book is a comprehensive guide to all aspects on paediatric intensive care The fourth edition has been fully revised to include the latest guidelines and advances in technology The extensive text of 1200 pages explains practical and surgical issues with thorough coverage of respiratory and cardiac care Other conditions specific to different systems of the body are also discussed endocrine gastrointestinal neurological and more Several chapters are dedicated to environmental injuries including burns electric shock heat disorders near drowning and poisoning The book concludes with discussion on psychosocial issues ethical and medicolegal aspects training research quality improvement and use of therapeutic drugs in paediatric intensive care The text is highly illustrated with clinical photographs diagrams and flowcharts Key points Comprehensive guide to all aspects of paediatric intensive care Fully revised fourth edition featuring latest guidelines and technological advances Extensive text of 1200 pages further enhanced by clinical photographs diagrams and flowcharts Previous edition 9789351527398 published in 2015

Respiratory Care: Patient Assessment and Care Plan Development David C. Shelledy, Jay I. Peters, 2016 For all students and clinicians assessing or caring for patients with cardiopulmonary disorders Respiratory Care Patient Assessment and Care Plan Development is a must have resource As the most comprehensive reference available it is a guide to the evaluation of the patient and the development and

implementation of an appropriate evidence based respiratory care plan Respiratory Care Patient Assessment and Care Plan Development describes the purpose of patient assessment and then guides the reader through the process of the reviewing existing data in the medical record conducting the patient interview performing the physical assessment and finally evaluating the diagnostic studies needed and implementing a respiratory care plan Bridging the gap between patient assessment and treatment the reader will learn how to apply assessment skills to the development and implementation of respiratory care plans Integrated throughout each chapter are Clinical Focus exercises RC Insights and Key Points to help readers refine critical thinking and problem solving skills as well as strongly grasp important concepts Chapter 1 Introduction to Patient Assessment Chapter 2 Development and Implementation of Respiratory Care Plans Chapter 3 Review of the Medical Record Chapter 4 Patient History Chapter 5 Physical Assessment Chapter 6 Assessment of Oxygenation Chapter 7 Assessment of Ventilation Chapter 8 Blood Gas Analysis Hemoximetry and Acid Base Balance Chapter 9 Laboratory Studies Chapter 10 Cardiac Assessment and the Electrocardiogram Chapter 11 Cardiopulmonary Imaging Chapter 12 Adult Pulmonary Function Chapter 13 Bronchoscopy and Special Procedures Chapter 14 Acute and Critical Care Monitoring and Assessment Chapter 15 Obstructive Sleep Apnea Chapter 16 Neonatal and Pediatric Assessment

The Integrated Nervous System Walter J. Hendelman, Peter Humphreys, Christopher R. Skinner, 2017-07-12 This innovative textbook is modelled on problem based learning It bridges the gap between academic neuroanatomy and clinical neurology and effectively takes the reader from the classroom to the clinic so that learning can be applied in practice This second edition has been updated and expanded to include many more clinical cases within both the book and the accompanying Web site This book and the associated Web site will be of practical value to all the professionals who deal with people who have neurological conditions as well as being invaluable to medical students and residents This includes psychiatrists rehabilitation medicine specialists physiotherapists occupational therapists and speech therapists and nurses who specialize in the care of neurological patients We think that this text will also be of value for family physicians and specialists in internal medicine and pediatrics all of whom must differentiate between organic pathology of the nervous system and other conditions

Pediatric Pelvic and Proximal Femoral Osteotomies Reggie C. Hamdy, Neil Saran, 2018-10-04 This unique case based text offers a comprehensive discussion of pelvic and proximal femoral osteotomies in the pediatric population Beginning with chapters on preoperative planning and radiologic evaluation of the adolescent hip subsequent chapters are sensibly divided into three thematic sections which use a consistent chapter format presenting the case history relevant imaging treatment goals the management strategy and clinical pearls and pitfalls Part I describes the various pediatric pelvic osteotomies including the Salter Pol de Coeur T nnis Pemberton and San Diego approaches among others Pediatric proximal femoral osteotomies comprise part II presenting the McHale procedure varus and valgus osteotomies Morscher osteotomy and Shepherd s Crook deformity to name just a few The final section covers combined and miscellaneous osteotomies and

procedures for the pediatric hip such as osteochondroplasty hip instability hip arthrodesis and SUPERhip and SUPERhip2 procedures for congenital femoral deficiency Each chapter is generously illustrated and includes a handy table of indications and contraindications for the procedure described In infancy childhood and adolescence the hip joint is very susceptible to abnormalities congenital or acquired that may lead to morphological alterations with potential sequelae specifically pain and difficulty to ambulate sit and perform daily activities Restoring normal anatomy and biomechanics of the hip joint by various pelvic and or proximal femoral osteotomies remains the cornerstone in the management of these conditions To this end Pediatric Pelvic and Proximal Femoral Osteotomies will be an invaluable resource for all pediatric orthopedic surgeons trainees and students both in the medical and paramedical field

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