Neuromuscular Disease: A Case-Based Approach

Neuromuscular Disease

A Case-Based Approach

Second Edition

Jessica E. Hoogendijk
University Medical Center Utrecht

Marianne de Visser
Amsterdam University Medical Center

Pieter A. van Doorn
Erasmus MC, University Medical Center Rotterdam

Erik H. Niks
Leiden University Medical Center

CAMBRIDGE UNIVERSITY PRESS

Shaftesbury Road, Cambridge CB2 8EA, United Kingdom

One Liberty Plaza, 20th Floor, New York, NY 10006, USA

477 Williamstown Road, Port Melbourne, VIC 3207, Australia

314–321, 3rd Floor, Plot 3, Splendor Forum, Jasola District Centre,
New Delhi – 110025, India

103 Penang Road, #05–06/07, Visioncrest Commercial, Singapore 238467

Cambridge University Press is part of Cambridge University Press & Assessment,
a department of the University of Cambridge.

We share the University's mission to contribute to society through the pursuit of
education, learning and research at the highest international levels of excellence.

www.cambridge.org
Information on this title: www.cambridge.org/9781108744188

DOI: 10.1017/9781108881593

When citing this work, please include a reference to the DOI 10.1017/9781108881593

First published 2013
Second Edition 2025

Printed in the United Kingdom by CPI Group Ltd, Croydon, CR0 4YY

A catalogue record for this publication is available from the British Library.

Library of Congress Cataloging-in-Publication Data
Names: Hoogendijk, Jessica E., 1957– author. | Visser, Marianne de, author.| Doorn, Pieter A. van, author. | Niks, Erik H., author.
Title: Neuromuscular disease : a case-based approach / Jessica E. Hoogendijk, University Medical Center Utrecht, Marianne de Visser, Amsterdam University Medical Center, Pieter A. van Doorn, Erasmus MC, University Medical Center Rotterdam, Erik H. Niks, Leiden University Medical Center.
Description: 2nd edition. | Cambridge, United Kingdom ; New York, NY : Cambridge University Press, 2024. | Revised edition of: Neuromuscular disease / John H.J. Wokke [et al.]. 2013. | Includes bibliographical references and index.
Identifiers: LCCN 2024017717 | ISBN 9781108744188 (paperback) | ISBN 9781108881593 (ebook)
Subjects: LCSH: Neuromuscular diseases – Case studies. | Musculoskeletal system – Diseases – Case studies. | Diagnosis, Differential – Case studies.
Classification: LCC RC925.55 .H66 2024 | DDC 616.7/44–dc23/eng/20240509
LC record available at https://lccn.loc.gov/2024017717

ISBN 978-1-108-74418-8 Paperback
ISBN 978-1-108-88159-3 Cambridge Core

Every effort has been made in preparing this book to provide accurate and up-to-date information that is in accord with accepted standards and practice at the time of publication. Although case histories are drawn from actual cases, every effort has been made to disguise the identities of the individuals involved. Nevertheless, the authors, editors, and publishers can make no warranties that the information contained herein is totally free from error, not least because clinical standards are constantly changing through research and regulation. The authors, editors, and publishers therefore disclaim all liability for direct or consequential damages resulting from the use of material contained in this book. Readers are strongly advised to pay careful attention to information provided by the manufacturer of any drugs or equipment that they plan to use.

Contents

Contents

Disorders of the Neuromuscular Junction

Myopathies

Supplementary videos are available at www.cambridge.org/neuromuscular

Contributors

Pieter van Doorn
Department of Neurology
Erasmus MC, University Medical Center
Rotterdam, The Netherlands

Stephan Goedee
Department of Neurology
University Medical Center Utrecht
Utrecht, The Netherlands

Jessica Hoogendijk
Department of Neurology
University Medical Center Utrecht
Utrecht, The Netherlands

Erik Niks
Department of Neurology
Leiden University Medical Center
Leiden, The Netherlands

Wouter van Rheenen
Department of Neurology
University Medical Center Utrecht
Utrecht, The Netherlands

Marianne de Visser
Department of Neurology
Amsterdam University Medical Center
Amsterdam, The Netherlands

Renske Wadman
Department of Neurology
University Medical Center Utrecht
Utrecht, The Netherlands

Foreword

During the past three decades, interest in neuromuscular medicine has increased exponentially.

This neurology section turned from a descriptive diagnostic setting into a molecular diagnostic and currently gene therapeutic subject.

However, the base of all this development remains the deep phenotyping or, more holistically, the Gestalt approach to neuromuscular disorders.

In this book, four 'icons' of the neuromuscular world, Marianne de Visser, Jessica Hoogendijk, Erik Niks, and Pieter van Doorn, jointly describe emblematic clinical case reports of the whole spectrum of these acquired and inherited more than 800 distinct disorders.

In this book, all sections of the nervous system, from motor neurons via the spinal cord and peripheral nerve through the neuromuscular endplate into the skeletal muscle, are reflected in case presentation to raise awareness, interest, knowledge, and clues for beginners, professionals, and to esteemed experts of the field.

A state-of-the-art summary supports these case reports for evaluating people with neuromuscular disorders using neuromuscular-specific ancillary investigations such as electrophysiology, imaging, and genetic testing by Stephan Goedee and Wouter van Rheenen.

This book, written in the beautiful Dutch tradition of clinical perception of medicine, will help set the framework for teaching and learning about patients living with neuromuscular disorders.

Taking up this ongoing novel 'therapy area' of neuromuscular disorders, this book is in high demand, as treating rare diseases is re-learning rare diseases.

Benedikt Schoser

Preface to 2nd Edition

Neuromuscular diseases (NMD) are classified as rare diseases affecting less than 5 individuals in 10,000, but collectively NMD are quite common. Diagnosis of NMD can be difficult, but due to advancements in particular in genetic testing, some adult patients may receive a better-defined or alternative diagnosis to the one established during childhood.

This book contains 66 neuromuscular cases across the spectrum of NMD that aim to familiarize the reader with the wide spectrum of clinical features in NMD, the diagnostic strategies, and therapeutic possibilities. Asking about symptoms and problems in daily life, the family history, and careful neurological examination often largely help to make an appropriate clinical and differential diagnosis. In the second edition of this book, 23 tables of differential diagnoses are included that assist in the diagnostic process. Additional laboratory investigations are often helpful and include at least assessment of serum creatine kinase (CK) activity in patients suspected to have a myopathy, anti-acetylcholine receptor, and other antibodies in patients with fluctuating weakness, and a range of laboratory tests to identify risk factors for polyneuropathy in patients with a neuropathy. The basics of electromyography (EMG) and nerve ultrasound (NUS) are described and illustrated. EMG examination is important in the diagnosis of motor neuron disease, helpful to diagnose polyneuropathy, and essential to dissect polyneuropathy into axonal and demyelinating subgroups. If a hereditary disease is suspected, up-to-date DNA analysis is often the diagnostic approach of choice. The basics of genetic diagnostic tests, including suggestions when and how to use them, are described. In inflammatory diseases of muscle, myositis blots to detect myositis-specific antibodies can be used to search for specific comorbidities. Advanced imaging techniques, especially MRI and muscle ultrasound, can be helpful to make the diagnosis or to guide the location of a muscle biopsy. An appropriate sequence of examinations usually allows for making an accurate diagnosis. However, despite advanced and targeted investigations, offering a precise diagnosis may remain a challenge.

Once a diagnosis has been made, other medical disciplines sometimes have to be consulted, for example a medical geneticist in case of a hereditary disease. Some patients with a hereditary myopathy, but also those with, for example, amyloidosis, require consultation of a cardiologist. Targeted treatment for hereditary neuromuscular disorders is currently limited to only a few diseases, but it is hoped that gene therapy will become more widely applicable in the near future. Most patients with immune-mediated diseases, especially those with immune-mediated polyneuropathy, and patients with myasthenia gravis or myositis can nowadays be treated successfully, but there is still a need for more effective drugs with fewer side effects, and new drugs are currently under investigation.

Management of patients with an NMD often also involves rehabilitation care in all its aspects, and there are current standards of care for anaesthesia, pregnancy, the transition of paediatric care to adulthood care, and palliation. Therefore, this book is relevant for a wide range of medical professionals, care givers, students, and others interested in patients with an NMD.

We are indebted to the late Professor John Wokke for his eminent contribution to the first version of this book, which was published in 2013. This second edition is fully revised and updated, now containing also paediatric cases and many new cases, new figures, and videos. To assist in the diagnostic process, we added 23 tables of differential diagnoses of a wide range of clinical problems (e.g., axial weakness, bulbar weakness, or pure motor distal weakness). However, we did not aim at completeness and this book is not intended to be a comprehensive textbook.

We thank Drs Stephan Goedee (Chapters 4 and 5), Wouter van Rheenen (Chapter 7), and Renske Wadman (Case 7) for their contributions, Professor

Eleonora Aronica and Dr Wim van Hecke for their help with the muscle pathology, Professor Peter Barth and Dr Anneke van der Kooi for providing Cases 58 and 64 Dr Janneke Hoeijmakers and Professor Jan Verschuuren for providing Figures 14.3 and 32.1, Professor Karin Faber and Dr Henk-Jan Westeneng for providing Videos 39.2-3 and 62.1, and Dr Merel van Maerle for her recommendations regarding genetic counseling in Chapter 8.

We foremost thank our patients, who form the basis of this book. They agreed to have their disease, or their child's disease, pictured in a photograph or video, trusting this will help in educating medical professionals and thus contribute to the treatment and care for patients with a neuromuscular disease.

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