Disorders of Voluntary Muscle
Disorders of Voluntary Muscle

Eighth edition

Edited by
George Karpati
David Hilton-Jones
Kate Bushby
Robert C. Griggs
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On-line Updates

As part of the modernization of this leading textbook, regular update bulletins on each chapter will be published on the website: www.cambridge.org/Karpati

On-line Updates will be authored by a team of outstanding neuromuscular disease specialists who do not currently contribute to the book. The updates will be published every six months, starting in June 2010 and will include selected new references. All content will be peer-reviewed by the Editorial team prior to release on the website.

We hope that this service will be of value to readers. The Editors and Publisher would welcome your feedback.
Foreword by John Walton (Lord Walton of Detchant)

I can hardly believe that this book is now entering its eighth edition. As I said in my foreword to the seventh, it was about 57 years ago when I first began to work on diseases of muscle at the request of the late Professor F.J. Nattrass of Newcastle upon Tyne. During the first few years I endeavoured to identify all of the patients with neuromuscular disease in its many varieties in the northeast of England, and this work led eventually to the introduction of a new classification of the muscular dystrophies, published by Nattrass and myself in Brain in 1954. I was fortunate to be able to spend a year learning neuropathology and the pathology of muscle from Raymond Adams at the Massachusetts General Hospital in Boston between 1953 and 1954, before spending a year in the Neurological Research Unit at the National Hospital Queen Square in London, where I continued with my clinical research. Our paper in Brain resulted in my receiving a substantial research grant from the Muscular Dystrophy Association of America, which enabled me to expand my research programme when I eventually returned to Newcastle in 1955. Later, with further grants from the Muscular Dystrophy Association of Canada and the embryo Muscular Dystrophy Group of Great Britain and Northern Ireland (which Nattrass and I founded in the early 1950s), I was able to embark upon a much expanded programme, for the first time involving basic research into neuropathology, histochemistry, electrophysiology and the biochemical aspects of neuromuscular disease, among other techniques of investigation. I had also developed the first service in electromyography and related techniques in Newcastle. Later still, with the aid of a programme grant from the Medical Research Council and support from the Wellcome Trust, among other charitable organizations, we were able to build a major research unit in the privately funded laboratories adjacent to the Regional Neurological Centre in Newcastle.

I presume that it was because of these developments that I was invited in 1962, by Mr. J.A. Rivers, of J&A Churchill Ltd, to edit a comprehensive volume on disorders of voluntary muscle, embracing basic science, clinical investigative techniques, clinical diagnosis and genetics, among other disciplines. Thus was Disorders of Voluntary Muscle born, and I was delighted to be able, as knowledge expanded at a remarkable rate, to see the book through five subsequent editions. In 1994, however, I recognized that, as I had passed my 72nd birthday and was not involved directly in clinical and laboratory research, or indeed in clinical practice, it was no longer appropriate for me to edit this volume, and was delighted when George Karpati of Montreal, David Hilton-Jones of Oxford and Robert C. Griggs of Rochester, New York, agreed to take it on. It was under their skilled and innovative editorship that the seventh edition appeared in 2001 and proved, in my opinion and in that of many others, to be the most outstanding textbook on diseases of muscle then available. But even since 2001, the virtual explosion of knowledge in molecular biology and other related techniques, and indeed in methods of investigation and management of muscle disease, has meant that a new edition was essential if readers were to be able to consult an authoritative source on such recent developments.

I am delighted that the editors have chosen Professor Kate Bushby of Newcastle to join their team, in view of her outstanding contributions to the field and her distinguished membership of the team of investigators and clinical collaborators now working in Newcastle, partly in a joint Medical Research Council unit, created jointly between the University of Newcastle and University College, London.

This new volume has been remarkably well designed and constructed, the first section dealing with the scientific basis of muscle disease, and the second with methods of investigation. The editors themselves present in Section 3A outstanding descriptions of clinical assessment and a guide to classification, and the principles of prevention, management and treatment, while the extensive Section 3B deals with individual muscle diseases in comprehensive detail. Naturally, because of my involvement in the birth and subsequent lusty development of this volume, I look upon the emergence of a fascinating and comprehensive eighth edition with a mixture of avuncular, even paternal, pride and pleasure. The editors have done a magnificent job in providing a volume which will stand as an outstandingly comprehensive guide to anyone interested in muscle in health or disease, whether basic scientist, clinical scientist, caring doctor or other healthcare professional: it will be read with pleasure and profit, to the ultimate benefit of patients whose future, because of massive developments in the last few years, is so much brighter than it was when the book originally appeared all those years ago.
Addendum

After I had completed this Foreword I learnt the devastating news of the sudden, untimely, and unexpected death of the principal editor of this volume, my good friend George Karpati. Without question, every doctor or scientist working in the field of neuromuscular disease in all parts of the world will be familiar with and will have admired the outstanding contributions which George has made to our understanding of the clinical and scientific aspects of neuromuscular disease throughout his distinguished professional lifetime. Hungarian by birth, George nevertheless became a proud and adopted Canadian, and his department in Montreal acted as a magnet to researchers and interested clinicians from across the world. So much more could be said, and no doubt will be in obituary notices, but speaking for myself I can only say that I have lost a dear and valued friend, whose wise counsel and comment at innumerable scientific meetings has always been to me a source of continuing edification and admiration. He has left a mark upon the field of neuromuscular disease which can never be erased, and will be deeply mourned throughout the scientific world. I shall remember him with pleasure and affection, and hope that this edition of Disorders of Voluntary Muscle will stand as an appropriate tribute to his contributions and to his memory.

John Walton (Lord Walton of Detchant)
Belford, Northumberland
June 2009
Myology as a discipline has continued to expand and increase in complexity since the previous edition of this book appeared in 2001. This growth has been due, mainly, to the application of molecular science to the field, which has led to the discoveries of new entities, a better understanding of the pathogenesis of the relevant diseases, improved diagnostic approaches, and a surge of advanced treatments. The editors have made every effort to ensure that the eighth edition of Disorders of Voluntary Muscle reflects these advances. This has been achieved by adding new chapters and by expanding the authorship; however, maintaining a manageable size necessitated condensing and combining chapters. Our ultimate aim is to provide the reader with an up-to-date, authoritative text that will facilitate patient care. Therefore, the authors have concentrated on practical aspects of muscle diseases supported by the use of first class illustrations. While the scientific basis of muscle disease has been addressed, we believe that for more detailed scientific aspects of muscle biology, the reader can consult appropriate reference books and journal articles.

In order to keep abreast of new developments in the future, we have introduced an on-line supplementary section [www.cambridge.org/Karpati] in which additional information and illustrations will be periodically generated, mainly by rising stars of myology.

The editors and the publisher welcome Dr. Kate Bushby of Newcastle upon Tyne, UK as a new editor. She brings vast experience and wisdom to the editorial process. Lord Walton’s contribution of a new Foreword remains a valuable nostalgic feature of the book.

The editors wish to thank the contributing authors for their expert contributions and the publisher for expediting timely publication.

Ever since the Disorders of Voluntary Muscle was first published by John Walton in 1964, it has been considered as the leading comprehensive clinical resource in myology. The editors are confident that this preeminent role will continue with the publication of the eighth edition.
George Karpati, senior editor of this textbook and leading molecular myologist and experimental neuropathologist of our generation, died suddenly February 7, 2009. George possessed the outstanding skills of a clinical neurologist, an experimental neuropathologist, and a molecular biologist. George’s monumental contributions to neuromuscular disease include his seminal studies of inclusion body myositis, critical illness myopathy, Duchenne muscular dystrophy, and carnitine deficiency. He first showed the localization of dystrophin to the muscle fiber surface in Duchenne dystrophy and demonstrated success with dystrophin gene replacement. Over the past two decades Dr. Karpati has been on the forefront of research on the molecular pathogenesis of muscle disease and he has become a dominant figure in approaches to the gene and cellular treatments of first animal models and then on to developing human trials of gene therapy for muscular dystrophy. He has received the highest level of recognition in Canada and abroad. He trained 30 research fellows now in leadership positions in Canada, the USA and around the world. His many awards included the Distinguished Scientist Award, Canadian Society of Clinical Investigation, 1997; Fellow of the Royal Society of Canada, 1999; Officer of the Order of Canada, 2001; Chevalier of the Order of Quebec, 2005; Member of the Canadian Academy of Health Sciences, 2005; Recipient of Prix du Québec, 2006; and Lifetime Achievement Award, World Federation of Neurology Congress, 2006.

George had finished coordinating and overseeing the editing of virtually this entire text at the time of his death. All three remaining editors knew George personally as well as professionally. We all had immense admiration for George’s creativity, energy, intensity, tenacity, and enthusiasm. We had all experienced first-hand his relentless pursuit of answers to the pathogenesis of the diseases that are his and our lives’ work. George is survived by his wife, Shira, and his two sons. George’s family, friends, and all of clinical neuroscience have suffered a great loss. We dedicate this book to our friend: George Karpati.

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