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Book review

Genetic Neuromuscular Disorders: A Case-Based Approach 2nd Edition Corrado Angelini. Springer Softcover £109.99, eBook £87.50

In this book Angelini proposes to bring to life the breadth of genetic advances in neuromuscular disorders for neuromuscular physicians and neurology residents through an illustrative case-based approach. Angelini's extensive personal experience and knowledge of neuromuscular disease comes across in this book through the number of personal cases, many of them published, referenced throughout. The book is organised into five sections including Muscular Dystrophies, Congenital Myopathies, Ion Channel Disorders, Metabolic Myopathies (Mitochondrial Myopathies are included here) and Neurogenic disorders. Classification systems for neuromuscular disorders are usually based on either phenotype or genotype i.e. clinical features or causative gene. Within each section, Angelini has chosen to present entries according to clinical classification and in keeping with the Online Mendelian Inheritance in Man (OMIM) system of disease name, gene name, OMIM number etc. For each condition there is a brief description, illustrative case(s) followed by laboratory findings, conclusion, key points and references. Many of the illustrative cases also include helpful patient photographs and illuminating histopathological images.

The book assumes a considerable amount of general knowledge about neuromuscular disorders, as there is no introduction to the book to lay out an overview of presenting symptoms of neuromuscular disorders, pertinent complications that should be actively investigated, such as cardiac and respiratory involvement, nor an algorithm for investigation when neuromuscular disease is suspected. Neither is there a sub-introduction for each subsection of the book, which results in much repetition in many of the descriptions of related disorders.

The absence of a general introduction makes this book less accessible to non-neuromuscular specialists, although Angelini says this is not his target audience. A concise introduction, however, I believe would still be of interest, even to the specialist practitioner and in addition inclusion of a schematic diagram illustrating the location of proteins from the extracellular matrix, the sarcolemma, the sarcomere, the cytosol and the nucleus, in muscle that are either deficient or dysfunctional in the muscular dystrophies or congenital myopathies presented would help bring together an overview of how all these proteins work together in concert in normal muscle function. Similar concise introductions might benefit other subsections.

Throughout the book there is use of some words that may have been lost in translation to English such as the use of the word prosthesis instead of orthotic.

Overall this is a comprehensive overview of genetic neuromuscular disease with only the occasional omission (eg TRPV4 related neurogenic disorders) and a wealth of "real" rather than textbook cases impressively illustrate the range of presentations for many of the disorders.

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