Neurogenetics (What Do I Do Now)

Christine MD Klein, Kishore R. MBBS, FRACP Kumar, Carolyn M. MBBS, FRACP, PhD Sue, Alexander Prof Dr Med M

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Aims
To some, the field of neurogenetics appears perplexing and indecipherable. In this volume, we will address this issue by providing clinicians with a framework for dealing with these disorders. This book is not intended to be an in-depth, comprehensive review of all neurogenetic conditions from 'A to Z'. Instead, we will provide a concise discussion using case studies to illustrate the most important and topical neurogenetic disorders. This case-based approach will make the book easy to reference, clinically relevant, approachable, and, we feel, more interesting.

Scope
The contribution of genetics to many neurological diseases is becoming increasingly apparent, and so it is imperative to stay up-to-date with these conditions. The 31 chapters in this volume cover a wide range of inherited conditions including forms of dystonia, Parkinson disease, spastic paraplegias, mitochondrial diseases, myopathies, neuropathies, and much more. Particular attention is paid to practical issues regarding how to make a genetic diagnosis and how to counsel the family. We will also address some contemporary issues in neurogenetics, such as the impact of direct-to-consumer genetic testing.

General Approach
In keeping with the WDIDN series, each chapter commences with a brief case study, which will be used as an example of an important condition in neurogenetics. The discussion will then be centered on the case, with a focus on crucial issues regarding the clinical assessment, investigations and management of these conditions. Key clinical points will be listed at the end of the chapter, along with a list of suggested further reading. All case studies in this book are based on real patients seen by the authors or their colleagues.

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