BOOK REVIEW

A book to learn essentials of classic clinical genetics in the area of genomics

New Clinical Genetics - 3rd edition
Edited by: Andrew Read and Dian Donnai
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Eight years after the first edition of the popular clinical textbook by Read and Donnai, the next revision is now available on the shelves of the stores. The authors are known highly professional experts of medical genetics, both of them are recognised scientists worldwide as well. Andrew Read also has expertise in human molecular genetics, whereas Dian Donnai is an exceptional dysmorphologist and syndrome specialist, so their co-authorship could cover all major areas of the contemporary clinical genetics—alloying the traditional areas and the new fields.

As the authors state in their preface, much has changed in the few short years after the second edition, so they had to revise and update almost every chapter of the textbook. The major driving force, the biggest single change, has been the adoption of the next-generation or massive parallel sequencing in the routine diagnostics. This indeed changed the service that can be offered by the geneticists to the families affected by rare and undiagnosed diseases. At the same time,

some approaches became less important, so the authors tried to move the balance more towards sophisticated techniques. The role of the clinical geneticists is evolving, the developments have mandated changes throughout the entire book, with special emphasis in Chapter 5, where the authors explain to the readers how sequencing works and is used, how it can be employed in the sensitive communication with families, particularly with affected individuals, or with those members who are at risk but as yet unaffected.

Beside this breakthrough, attention was devoted to revise 'Disease Boxes' on mosaicism, RASopathies and disorders of the spliceosome in Chapter 7; emphasis on 'omic' methods in many places, particularly in Chapter 12 on cancer. The references are also well updated.

This is a good book, it is comprehensive and very useful. It can be recommended primarily to undergraduates and postgraduates in medicine, including the board exam trainees, and to PhD students working on human disease genetics, who can profit from reading it. It can also be recommended to those who work in laboratory genetics and want to improve their basic knowledge in clinical genetics.

CONFLICT OF INTEREST

The author declares no conflict of interest.

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