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BOOK REVIEW

The practical implications when finding chromosome abnormalities

'Chromosome Abnormalities and Genetic Counselling'. 4th Edition, Edited by RJ McKinlay Gardner, Grant R Sutherland and Lisa G Shaffer
ISBN: 978-0-19-537533-6
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The fourth edition of this reference book has been extensively revised to include the new technical developments in cytogenetics that have had advanced our understanding of the underlying genetic mechanisms and disorders. This text book continues to serve its main objectives well/admirably and lays the foundations for good practice in the analysis, interpretation and explanation of cytogenetic abnormalities for both clinical geneticists and laboratories.

Since the publication of the third edition in 2004, there have been major developments in the field of genetics with the introduction of more molecular-based techniques. The traditional boundaries between molecular genetics and cytogenetics are now disappearing with the advent of microarrays and next-generation sequencing (NGS) coupled with more precise knowledge of the gene content of each chromosome. Consequently many chapters have been updated as well as restructured to incorporates additional data obtained using these new technologies including some new chromosomal syndromes. In addition, two new chapters have been added: Chromosome Analysis, which includes microarray, QF-PRC, MLPA and next-generation sequencing techniques, and a chapter on Copy Number Changes that includes definitions, normal population variants, prenatal diagnosis and results of unclear clinical significance. Topics such as Robertsonian fission, Fragile X pre-mutation carriers, the new nomenclature for sex development disorders as well as a broader coverage of prenatal diagnostic tests are now incorporated into this fourth edition. The book has also benefited from the addition of some colour plates and the revision of some of the other figures present in the third edition. The literature references include the latest updates (as far is possible in a text book).

Much of cytogenetics relies on the empirical knowledge as well as experience in the field. The authors have ensured that each chapter has a section covering the genetic counselling issues so that clear, comprehensive and balanced advice is given.

This book will continue to be an indispensable tool for both clinical geneticists and laboratories because of the comprehensive and educational nature of its content■

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