



Chromosome Abnormalities and Genetic Counseling (Oxford Monographs on Medical Genetics)

R. J. M. Gardner, G. R. Sutherland

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There have been many advances in clinical cytogenetics since the first edition of this book appeared in 1989. The authors have written more expansively on segregation and risks in reciprocal translocations, X-autosome translocations, inversions, insertions, and prenatal diagnosis. The deletion syndromes have become more numerous, and more precisely defined, and this required treatment. Fluorescence in situ hybridization has become a routinely applied methodology, and its use has extended the power, and increased the sophistication, of the discipline of clinical cytogenetics. The Human Genome Project is unravelling the complexity of our genetic inheritance, and readers will find evidence of its impact on the practicalities of human cytogenetics throughout the book. Two phrases not used at all in the first edition are dynamic mutation and genomic imprinting. The fragile X syndrome now has the status of the prototypical dynamic mutation. The authors completely rewrote this chapter, giving it a much more molecular character. The Prader-Willi and Angelman syndromes, likewise, have become the classic examples of genomic imprinting and uniparental disomy. Compared with the tentative comments on these syndromes in the first edition, they now receive more extensive treatment. Yet, while the subject has become more complicated, the book's aim remains the same; to furnish a straightforward scientific description that will help readers understand the various chromosome abnormalities encountered in clinical practice, and to provide practical advice that can be passed on to the people who have, or whose families have, these abnormalities.

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