Book Review

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The Principles of Clinical Cytogenetics, second edition. Edited by Steven L. Gersen and Martha B. Keagle. Totowa, NJ: Humana Press, 2004. Pp. 596. $145.

The Principles of Clinical Cytogenetics is an essential reference book for all clinical cytogeneticists. It is written with a unique laboratory focus; it answers the most frequently asked questions about chromosomes and chromosomal disease and presents a comprehensive look at the field as it stands today. Updated after 5 years by many of the original authors, most of whom are practicing laboratory cytogeneticists, it includes sections on basic concepts, chromosome analysis, clinical cytogenetics, cancer cytogenetics, FISH, and even a section, entitled "Beyond Chromosomes," that covers DNA analysis of fragile-X syndrome, imprinting, uniparental disomy, and counseling. The book is clearly written and readable, aimed at the career cytogeneticist (technicians, supervisors, and directors), clinical staff involved in the day-to-day operation/interpretation of laboratory data, genetic counselors, medical students, fellows, and even undergraduates rotating through the lab. A careful reading of this book would serve as excellent review for the American Board of Medical Genetics examination. An e-book is included with every copy.

Although the cover is still easily recognizable and many of the chapters have their original authors, the basic information in the second edition has been elegantly revised, with the inclusion of new entries on infertility and chromosome instability and an expansion of the cancer cytogenetics and FISH sections. The editors even apologize for not holding up publication to include the latest International System for Cytogenetic Nomenclature (due out next year). I particularly enjoyed the discussions of chromosome abnormalities in light of recent information available from the Human Genome Initiative. For example, the chapter on structural chromosome rearrangements includes a discussion of the underlying molecular mechanisms that lead to recurrent deletion syndromes and the complementary duplications/triplications of those regions. The authors' introductions to each chapter lend cohesion to the book, perspective, and sometimes a bit of advice. Relevant clinical information is fairly easy to understand, with medical terms defined and clearly constructed tables included. The pictures and ideograms are of high quality, and the references at the end of each chapter are very comprehensive and will be helpful in the laboratory when a new chromosome abnormality is encountered. As a part of this review, I asked my laboratory staff each to read and report on a chapter at our lab meeting. This was a very helpful exercise in raising awareness, and it sparked productive discussion.

In its first 50 years of development, clinical cytogenetics has undergone significant change, starting with correct chromosome counting, followed by reproducible chromosome banding and FISH. We are now on the verge of yet another significant change in cytogenetics, as we begin to incorporate chromosomal microarrays into our laboratories. It is the perfect time to recap our progress and to consider how much we still have to learn to answer the basic questions about chromosome behavior and the mechanisms that lead to chromosomal abnormalities. I can only hope that when this text is updated to a third edition I will be around to read the book. At this moment in time, Gersen and Keagle tell the story better than anyone.

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