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Book Review

MEDICAL GENETICS
Lynn B. Jorde,
John C. Carey, and Raymond L. White
St. Louis: Mosby-Year Book, Inc., 1995, pp. 293.

Fortunately, since 1990 a handful of affordable textbooks have been published that are specifically designed to provide students who have little or no background in medical genetics with a broad, intermediate knowledge base of general concepts in this subject. This particular medical genetics textbook, which was published in 1995, is clearly one of the most current and appealing of its genre. It nicely blends the collective wisdom of well-versed and well-respected medical and molecular geneticists from three different departments (J.C. Carey, Pediatrics; L.B. Jorde, Human Genetics; and R.L. White, Cellular, Viral, and Molecular Biology) at the University of Utah Health Sciences Center. Their talents in teaching basic concepts in human genetics are clearly apparent throughout the text. By explaining key points through interesting, illustrative examples the authors should help make learning the subject matter a relatively painless process for even the most fearful-of-science student. It should also ensure that this textbook will be welcomed into a diverse group of classroom situations. Thus, it will provide nursing, medical, and allied health educators with an excellent choice among a limited number of such texts to accompany their courses in basic medical or human genetics.

One unique potential advantage of using this particular textbook for educational purposes is the availability of a set of 35-mm companion teaching slides that contain many of the figures and tables of the text. These may be purchased from the publishers, thus facilitating, at a reasonable cost, the design of a virtually "ready-made" course in medical genetics if desired. The questions at the end of each chapter will help assess a student's grasp of a chapter's content. Easy to understand answers are provided for each question at the end of the book.

The textbook consists of 12 clearly written chapters. The early chapters contain appropriate general background material specifically geared toward students with essentially minimal previous exposure to the subject. Specifically, early chapters titled "Basic Cell Biology: Structure and Function of Genes and Chromosomes" (Chapter 2, 23 pages) and "Autosomal Dominant and Recessive Inheritance" (Chapter 4, 28 pages) provide brief, yet comprehensive, introductions to their subject matter. Obviously in only 23 pages of Chapter 2, of which approximately 50% of the total pages are devoted to diagrams, just elementary concepts of cell biology are presented. Included in this chapter are concise explanations of the key concepts important in medical genetics including DNA replication, transcription, translation, meiosis, and mitosis—all of which are essential for students to comprehend prior to their understanding of specific concepts in medical genetics. It should be anticipated that students who have previous undergraduate courses in cellular biology, molecular biology, or genetics may find these early chapters to be a very rudimentary review and they may have little, if any, challenge in digesting the presented material. For these students, further reading from the references listed at the end of the chapter (often other book chapters or review articles from 1990 to 1994) will provide more detailed information as desired. For those students with little background in cellular or molecular biology, however, these chapters provide an easy-to-understand necessary framework upon which their further knowledge in medical genetics can be based. The later chapters of the textbook are devoted to specific medical genetic topics such as "Cancer Genetics" (Chapter 9, 16 pages) and "Genetic Screening, Genetic Diagnosis, and Gene Therapy" (Chapter 11, 22 pages). These chapters will help familiarize students with many genetic mechanisms of human disease and hopefully spark the interest of a few to pursue further study of molecular and medical genetics. It is very likely that this book will appeal to a diverse group of students as it not too text dense and its content can easily be covered in a single course.

One particular strength of this textbook is its visual emphasis. With its many illustrations and numerous scattered case reports it will undoubtedly encourage many students to read ahead, especially since sections of highlighted (bold font, red ink) inset text seem to pop off the page into the eyes of the non-color blind reader. This highlighted text features concisely worded key summary concepts of the text and facilitates a rapid, easy overview of main points contained within each chapter. The text itself also has key words or terms printed in bold black ink; the vast majority of these words are also clearly defined in the glossary at the end of the text. The majority of pages within this textbook also contain an easy to follow, large, illustrations (yes, unfortunately, in those familiar shades of red, white, and black) or tables to highlight key portions of the text. There is one double-sided glossy page of color diagrams embedded within Chapter 5 "Sex-Linked and Mitochondrial Inheritance." The five color plates on this page include two illustrations and two single-color probe FISH photographs that are already adequately presented elsewhere in the book. One color plate depicts an illustration of unequal crossing over for protanomalous and deuteranomalous color vision that could have been easily and clearly depicted using black, white, and red color scheme. Thus, these illustrations are poor choices for the few available color plates and are redundant. Color plates depicting images where color is necessary or use-

ful to illustrate a point (such as multicolor FISH analysis, automated sequencing profiles, or clinical photographs) would have been better choices for the color plates. The authors do add, at the end of their text, 89 bar codes representing different clinically related photographs to be retrieved from the "Slice of Life Illustrations" that are reportedly available in several medical campus libraries. While this is a creative approach in the medical genetic textbook market, it leaves readers who do not have ready access to the Slice of Life (including myself, although I was assured that it could be made available at our Medical Library if required by a course) feeling like we have been left out. It would be more useful in future editions to include these photos in the textbook itself or, if that is cost-prohibitive, to make them available through InterNet services that are likely more accessible to many readers.

Another particular strength of this book, when compared to similar textbooks, is that it nicely blends basic scientific concepts with pertinent, interesting clinical correlations essentially from the beginning of the text onward. These sections are set aside from the main text in a boxed-in-pink background and often include patient photographs. For instance, within Chapter 2 (where the basic structure and function of cells, chromosomes, genes, and proteins are presented) is a two and a quarter page "Clinical Commentary." It is titled "Osteogenesis Imperfecta, an Inherited Clinical Disorder" and includes a table highlighting the clinical features of OI Types I–IV, a photograph and radiograph of a stillborn infant with Type II OI, and a diagram illustrating the process of collagen fibril formation. This clinical correlation immediately drives home the important clinical relevance of understanding the basic concepts presented in the text. In chapter 6 where cytogenetic concepts are presented, the clinical commentary is "XX Males, XY Females, and the Genetic Basis of Sex Determination," which certainly would have grabbed the attention of many students in my medical school class. This three quarter page commentary discusses and diagrams the pseudoautosomal region of the Y chromosome and addresses the role of the SRY locus in sex determination. In the last chapter, Chapter 12, "Clinical Genetics and Genetic Counseling," seven clinical commentaries, which range from general concepts in genetic counseling to specific genetic disorders, are included to augment the text. Obviously, these brief clinical correlations can only provide a very limited introduction to the molecular mechanisms of human disease but should suffice to whet the appetite of interested students.

The several strong features of this textbook clearly outweigh the apparent typographical and editing errors that can, unfortunately, be found scattered throughout this first edition. It will be important for those who use this text in their courses to correct these mistakes to avoid causing unnecessary frustrating confusion among students—a likely result, for instance, when, while reading page 237, students try to correlate a pedigree with a paragraph of text describing the pedigree that contains wrong individual identification numbers. There is certainly no need to make this introduction to Bayesian analysis problematic for students as many students will have enough difficulty understanding and applying Bayes' theorem even given the clearest of explanations. One could also fault the text for all the omissions that are characteristic of such a textbook that, by design, can only offer a relatively limited sampling from a huge menu of topics. For instance, in this textbook there is very little presented regarding the genetics of inborn errors of metabolism. However, like other textbooks of this genre it is not, nor was it intended to be, a comprehensive catalog of all genetic disorders or the molecular basis of all human disease. It is also not designed to be the type of textbook that students in their clinical clerkship years would likely consult to further their understanding of applied genetic concepts as related to their direct patient management. Ideally, these students should have already benefited from the information that is provided in this textbook through earlier experiences or courses. Although not a fault unique to this particular book, some information about the various genes and diseases presented in this textbook is already out-of-date. This reflects the unprecedented rapid pace of advances in medical and molecular genetics that, in a large part, was sparked by the development of the ongoing Human Genome Project. Certainly it should be expected that new developments in the field will occur at least as quickly. It is therefore essential that this textbook be regularly updated, preferably with new editions every 3 years, in order to be a leading textbook in the field.

In summary, the broadly ranging topics covered in this textbook are, for the most part, succinctly and clearly presented. It is well illustrated and richly enhanced by the inclusion of several clinical examples. It should foster the development of almost any student's interest in medical genetics. These features, along with its competitive pricing, make it an ideal textbook for medical, nursing, allied health professional, and genetic counseling students.

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