Book Reviews

A Clinical Guide to Inherited Metabolic Diseases, 2nd ed.

J. T. R. Clarke.

Cambridge University Press, Cambridge, UK. 2002. 289 pp. $40.00.

In the second edition of this introductory text, J. T. R. Clarke again masterfully succeeds in summarizing inherited metabolic diseases for the non-metabolic specialist. The new edition updates the latest research as well as diagnostic and therapeutic tools, while expanding the discussion of mitochondrial and genetic contributions to disease. Despite the significant expansion of the field in the last six years, *A Clinical Guide to Inherited Metabolic Diseases* retains its readily accessible format, making it suitable for reference in a clinical setting or as a review text.

This book consists of 10 concise chapters and 288 pages, only minimally longer than the first edition. Undoubtedly, for most clinicians and trainees, the patient with a potential inborn error of metabolism remains quite intimidating, a situation not helped by flashbacks to the all-too-quickly-forgotten principles of biochemistry and physiology. The goal of the book is to calm those fears by introducing basic concepts within a clinically oriented framework. Clarke succeeds in this goal, distilling a very broad, complex, and ever-expanding field into a usable, well-written text that contains extensive clinical examples and a multitude of diagrams and tables to facilitate rapid reference. As was the first edition, the second edition is a necessity for all trainees and professionals who care for infants and children, especially in inpatient settings.

An author writing a review is faced with a dilemma: whether to write a "cookbook" with only algorithms and tables that can be carried around—but that might miss the opportunity to provide basic concepts and thereby enable long-term understanding of mechanisms—or to provide a review of all knowledge in the field in encyclopedic form, risking that the text will remain on the shelf, unavailable to the clinician. Clarke is able to address this dilemma and balance the short-term need for readily available diagnostic and treatment guidelines, yet he provides enough theory and explanation to provide a groundwork for understanding the mechanisms.

The second edition begins with an excellent review of basic principles, covering metabolism, genetics (including mitochondrial inheritance), and a brief introduction to diagnosis and diagnostic dilemmas. The next six chapters are arranged according to clinical presentation, allowing for rapid review of a topic. Each chapter is introduced with a concise summary of the physiology of the normal and disease states, and well-constructed cartoons and simplified pathways further facilitate understanding of the underlying physiology of the derangement. Clarke does not attempt to provide a laundry list of diagnoses associated with a derangement; rather, he presents and discusses several detailed examples of specific diseases, utilizing extensive clinical information provided in tables, charts, illustrations, and photographs. Each chapter closes with a discussion of diagnosis and treatment options. Throughout the book, chapters have multiple subtitles, further enabling rapid reference, and are followed by a brief bibliography of essential references, selected so as not to overwhelm those who desire a more in-depth investigation. The text has a thorough, easy-to-use index as well as a comprehensive list of Web sites for parents and for further reading or investigation.

New to this edition is a chapter on newborn screening, which includes a discussion of tandem mass spectrometry, a very timely topic for those who care for neonates and children or for those who formulate public policy. The final two chapters of the text address diagnosis and treatment in greater detail, again covering the treatment of specific diseases in more detail.

This book is compact, portable, and very reasonably priced at $40. Because it is arranged by clinical presentation rather than specific disease, and contains a myriad of tables, cartoons, photographs, and charts, it successfully balances the clinical need for expediency and the long-term desire for understanding concepts. The text is an excellent value and would be most useful to all pediatric and genetic resi dency libraries, as well as to any student or professional interested in the diagnosis and care of patients with metabolic diseases.

KEITH K. VAUX
Fellow in Dysmorphology and Medical Genetics
Department of Pediatrics
University of California–San Diego
San Diego, CA 92103

DOI: 10.1093/jhered/esg048