J Inherit Metab Dis (2017) 40:165
DOI 10.1007/s10545-016-0008-0

BOOK REVIEW

Jean-Marie Saudubray, Matthias R. Baumgartner, John Walter (Eds.). Inborn metabolic diseases: diagnosis and treatment

2016, 658 pp (ISBN 978-3-66249771-5)

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Published online: 12 December 2016
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Inborn Metabolic Diseases: Diagnosis and Treatment remains one of the standard textbooks for professionals working in inborn metabolic medicine and biochemical genetics. For this 6th edition, edited by Jean-Marie Saudubray, Matthias Baumgartner, and John Walter, all chapters have been newly written and updated by internationally recognized experts on their subjects without e xpanding the book's size. Like the previous editions the book is divided into eleven 11 sections with a total of 43 chapters. The first section maintains the well-tried quartet of clinical approaches to inborn errors of metabolism in paediatric and adult medicine, diagnostic procedures and emergency treatment including excellent tables, diagrams and powerful algorithms. The following sections and chapters provide detailed information and recommendations for diagnosis, treatment and follow-up for the single inborn metabolic diseases in a profound, well structured, inventive format. The field of inborn errors of metabolism is rapidly evolving with new disease categories (e.g. cytoplasmatic tRNA synthetases and related factors of cytoplasmic protein synthesis). As the authors write in their preface, since the previous edition published in 2011, more than 300 'new' disorders have been described. Already going quickly through the table of contents and the updated and added tables and algorithms in chapters 1 and 2 reveals, how careful new disorders and their presentation have been included in the 6th edition. Also, diagrams, tables and figures were redesigned and, for the first time, coloured making them not only more attractive but also more informative, now highlighting the central paragraphs, disease groups and single disorders. Missing are only more and coloured pictures of clinical findings and a table concentrating the biochemical approach to recognition of inborn metabolic diseases (analogous to the clinical approach). However, adding this would have probably expanded the book to a less convenient size. Here, for specific needs the reader might inquire a metabolic atlas and further books in addition. Being a paediatrician subspecializing for metabolic medicine, writing the book review, this book will accompany my further training and daily work as the previous editions did. Therefore, I can recommend it as an indispensable constant companion for clinicians and biochemists in particular, but also to all specialists dealing with inborn errors of metabolism. In conclusion, this sixth edition of *Inborn Metabolic Diseases: Diagnosis and Treatment* gives an excellent and most up to date overview and deep insight on a rapidly evolving specialty with a steadily growing number of patients, most of them now growing-up and reaching adulthood, and is, therefore, informative for experts as well as for those who seek to become one.

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