Preface to the 6th edition

Inborn Metabolic Diseases: Diagnosis and Treatment, remains the standard textbook for professionals working in inherited metabolic medicine and biochemical genetics but it is also an essential resource for all specialties in this multidisciplinary field. The speciality of inherited metabolic disease is at the forefront of progress in medicine with new methods in metabolomics and genomics identifying the molecular basis for a growing number of conditions and syndromes that were previously unexplained. These powerful techniques allow us to link the clinical, biochemical and molecular characteristics of disorders and provide a basis for therapeutic interventions.

For this new edition all 43 chapters have been revised or newly written by authors with particular expertise in their subject areas. Since the previous edition published in 2011, two new categories of inborn errors of metabolism (IEM) and more than 300 ‘new’ disorders have been described, 85% presenting with predominantly neurological manifestations. The chapters that encompass these have been considerably extended, including those involving complex lipids (phospholipids, triglycerides, sphingolipids) and non-mitochondrial fatty acid homeostasis (including peroxisomal defects) (>60 disorders), congenital disorders of glycosylation (>90 disorders), purine metabolism (35 disorders), metal transport (>35 disorders), and disorders of oxidative phosphorylation (including mitochondrial transporters, iron-sulfur complex metabolism and mitochondrial tRNA synthetases) (>230 disorders). The newly described metabolic disorders affecting cytoplasmic tRNA synthetases and other factors related to cytoplasmic protein synthesis, transporters, channels and enzymes implicated in the logistics and regulation of the cell, challenge our current classification based on organelles and form a bridge between ‘classic’ metabolic diseases with metabolic markers and those caused by mutations in structural proteins without such markers, which are most often diagnosed by molecular techniques.

While this new edition highlights recent findings it continues to provide a comprehensive review of all IEM, with a particular focus on the clinical and biochemical approach to recognition, diagnosis and treatment at all ages. The clinical algorithms of chapters 1 and 2 incorporate both ‘old’ and ‘new’ disorders, and there are now more algorithms detailing neurological presentations. An updated listing of metabolic markers and profiles and a section on molecular techniques such as next generation sequencing and gene panels have been added. In order to keep the book to a reasonable size we have not included a chapter dedicated to newborn screening in this edition; instead this method of diagnosis is discussed for individual disorders in their relevant chapters.

As before, we continue to advocate referral to specialist centres for the diagnosis and treatment of inherited metabolic disorders. For countries in the European Union a list of such centres is compiled by the Society for the Study of Inborn Errors of Metabolism (SSIEM), while for the United States and Canada, Japan, Australia, South American and Middle East countries comparable lists are compiled by the American (SIMD), Japanese (JIMD), Australian (AIMD) South Latin America (SLEIMPN) and Middle East societies for the study of inherited metabolic diseases, respectively.

We pay tribute to our colleague George van den Berghe who has now retired from the editorial board. We welcome new authors and also thank those previous authors who, while not involved with this edition, have helped to lay the foundation for this book.

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