**Inborn metabolic diseases. Diagnosis and treatment**

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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 publication in 2011, two new categories of inborn errors or 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 affecting structural proteins without such markers and which are most often diagnosed by molecular technics.

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 technics 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.