



6th ed. 2016, Approx. 670 p. 81 illus., 69 illus. in color.

 **Printed book**

Hardcover

ISBN 978-3-662-49769-2

▶ **169,99 € | £126.50**

▶ ***181,89 € (D) | 186,99 € (A) | CHF 187.00**

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Inborn Metabolic Diseases

Diagnosis and Treatment

- ▶ **Essential resource in the interdisciplinary field of inborn metabolic diseases**
- ▶ **Indispensable for professionals in specialities ranging from pediatrics, neonatology, pathological biochemistry and genetics to neurology, internal medicine, nursing, dietetics and psychology**
- ▶ **Written by internationally recognised experts on their subjects**

This work is recognised as the standard textbook for professionals involved in the diagnosis and management of inborn errors of metabolism (IEM) and an essential resource in this multidisciplinary field. For the 6th edition all 43 chapters have been newly written or revised by authors with particular expertise in their subject areas.

Contents

A clinical and biochemical approach to the recognition and diagnosis of IEM with algorithms to symptoms, signs, and syndromes in patients of all ages; Emergency treatments; Medications.

Separate comprehensive sections on IEM of: Carbohydrates; Mitochondrial Energy; Amino and organic acids; Vitamin-responsive defects; Neurotransmitter and Small peptides, Lipid and Bile Acids; Nucleic Acid and Heme; Organelles.

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