Inborn Metabolic Diseases

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.
• Disorders affecting the synthesis and remodelling of complex lipids and fatty acid homeostasis are now included.

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