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Titolo	Inherited Metabolic Diseases : A Clinical Approach / / edited by Georg F. Hoffmann, Johannes Zschocke, William L. Nyhan
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ISBN	3-662-49410-8
Edizione	[2nd ed. 2017.]
Descrizione fisica	1 online resource (XVII, 605 p. 80 illus., 34 illus. in color.)
Disciplina	618.92
Soggetti	Pediatrics Internal medicine General practice (Medicine) Neurology Pathology Human genetics Internal Medicine General Practice / Family Medicine Neurology Human Genetics
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references at the end each chapters and index.
Nota di contenuto	Introduction to Inborn Errors of Metabolism: Disorders of Intermediary Metabolism -- Mitochondriopathies Neurotransmitter Defects.- Disorders of the Biosynthesis and Breakdown of Complex Molecules. Approach to the Patient: When to Suspect Metabolic Disease -- Patient Care and Treatment -- Metabolic Emergencies -- Anesthesia and Metabolic Disease -- Principles of Dietary Therapy -- Principles of Enzyme Replacement Therapy -- Principles of Gene Therapy. Organ Systems in Metabolic Disease: Cardiovascular Disease -- Liver Disease -- Gastrointestinal and General Abdominal Symptoms -- Kidney Disease and Electrolyte Disturbances -- Neurological Disease -- Metabolic Myopathies -- Psychiatric Disease -- Eye Disorder -- Skin and Hair Disorders -- Bone Disorders -- Physical Abnormalities in Metabolic Diseases -- Hematological Disorders -- Immunological

Problems. Investigations for Metabolic Diseases: Newborn Screening -- Biochemical Studies -- Enzymes, Metabolic Pathways, Flux Control Analysis and the Enzymology of Specific Groups of Inherited Metabolic Diseases -- Molecular Investigations (DNA Studies) -- Pathology / Biopsy -- Postmortem Investigations -- Neuroimaging -- Function Tests -- Suspected Mitochondrial Disorder. Appendix: Differential Diagnosis of Clinical and Biochemical Phenotypes -- Reference Books -- E3 Internet Resources.

Sommario/riassunto

This book focuses on clinical presentations that may be caused by inherited metabolic diseases. Its symptom- and system-based approach will help clinicians with and without detailed knowledge of human biochemistry in all specialties to reach a correct diagnosis and institute the optimal treatment program. The book summarizes the central elements of inherited metabolic diseases and describes clearly how to carry out an efficient yet complete diagnostic work-up, thereby guiding the clinician from the presenting symptoms and signs through to effective initial management. After an introduction to the different disorders, the book explains when to consider an inborn metabolic error and which initial tests to order. Core aspects such as structured communication, guidelines, transition, pregnancy, maternal care and how to respond to various medical emergencies are covered. Therapeutic concepts such as dietary treatment are delineated and practical advice provided on the quite different treatment approaches required for individual diseases. An extensive section structured according to organ systems outlines the correct approach in the context of specific symptoms and signs. The value of each of the potential investigations is explained, with precise advice on the interpretation of results. The inclusion of algorithms, tables, lists, and charts facilitates rapid decision making and information retrieval, and the appendices include a helpful guide to differential diagnosis based on clinical and biochemical phenotypes. This new updated edition of Inherited Metabolic Diseases will be an invaluable aid for the busy clinician and an excellent quick reference for metabolic and genetic specialists.