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Titolo	Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases / / edited by Nenad Blau, Marinus Duran, K Michael Gibson, Carlo Dionisi Vici
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Descrizione fisica	1 online resource (880 p.)
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Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and indexes.
Nota di contenuto	Introductory Chapters -- Amino acids -- Organic acids -- Vitamins and neurotransmitter -- Energy metabolism -- Organelles -- Selected disorder -- Biochemical phenotypes of questionable clinical significance -- Profiles.
Sommario/riassunto	This book, combining and updating two previous editions, is a unique source of information on the diagnosis, treatment, and follow-up of patients with inherited metabolic diseases. The clinical and laboratory data characteristic of the ever-growing number of rare metabolic conditions can be bewildering for the general clinician. Reference

laboratory data are scattered and clinical descriptions maybe obscure. The Physician’s Guide documents the features of more than five hundred conditions, grouped according to disorder category. Relevant clinical findings are provided and pathological values for diagnostic metabolites are provided. Signs and symptoms are provided for each disorder from birth through adulthood. In addition, the role of biochemical genetic testing is outlined. Treatment protocols and experimental therapies are fully described, with guidance on follow-up and monitoring. The authors are acknowledged experts from across the world, and the book will be invaluable to all who deal with patients with inherited metabolic diseases, including pediatricians, internists, neurologists, and clinical geneticists, as well as clinical and biochemical geneticists.