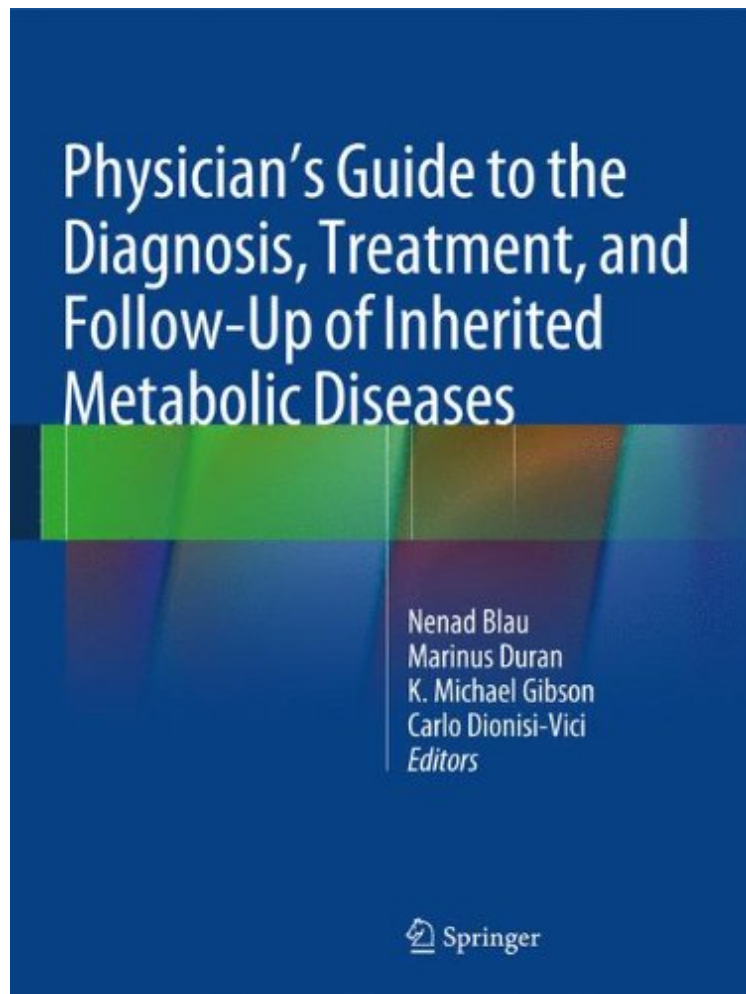


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## Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases

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This book, combining and updating two previous editions, is a unique source of information on the diagnosis, treatment, and follow-up of metabolic diseases. The clinical and laboratory data characteristic of rare metabolic

conditions can be bewildering for both clinicians and laboratory personnel. Reference laboratory data are scattered, and clinical descriptions may be obscure. The Physicians Guide documents the features of more than five hundred conditions, grouped according to type of disorder, organ system affected (e.g. liver, kidney, etc) or phenotype (e.g. neurological, hepatic, etc). Relevant clinical findings are provided and pathological values for diagnostic metabolites highlighted. Guidance on appropriate biochemical genetic testing is provided. Established experimental therapeutic protocols are described, with recommendations on follow-up and monitoring. The authors are acknowledged experts, and the book will be a valuable desk reference for all who deal with inherited metabolic diseases.

From the book reviews:It is a unique source of information on the diagnosis, treatment, and follow-up of patients with inherited metabolic diseases. much of the information is presented in tables which gives a good overview and makes the information easily readable. the book supplies clinicians and clinical biochemists with data that should facilitate the diagnosis and treatment of patients with inherited metabolic diseases. (Christian Staufner and Verena Peters, Journal of Inherited Metabolic Disease, Vol. 37, 2014)From the Back CoverThis 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 Physicians 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.