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 *Physician's 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.

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