



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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Editorial Review

Review

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 Cover

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.

Users Review

From reader reviews:

Julie Gailey:

Nowadays reading books become more and more than want or need but also get a life style. This reading behavior give you lot of advantages. The benefits you got of course the knowledge the actual information inside the book that will improve your knowledge and information. The information you get based on what kind of publication you read, if you want get more knowledge just go with training books but if you want experience happy read one together with theme for entertaining such as comic or novel. The actual Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases is kind of reserve which is giving the reader unstable experience.

Patricia Cockrell:

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

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Frederica Dawkins:

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