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Mark Steyvers

**Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases** Nenad Blau,Carlo Dionisi Vici,Carlos R. Ferreira,Christine Vianey-Saban,Clara D. M. van Karnebeek,2022-02-21 This updated and enlarged second edition 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 clinicians and laboratory personnel alike – reference laboratory data is scattered, and clinical descriptions can be obscure. The new Physician’s Guide with the additional more than 600 diseases now featured, documents 1200 conditions grouped according to type of disorder, organ system affected (e.g. liver, kidney, etc) or phenotype (e.g. neurological, hepatic, etc). It includes relevant clinical findings and highlights the pathological values for diagnostic metabolites. Guidance on appropriate biochemical genetic testing is also provided and established experimental therapeutic protocols are described, with recommendations on follow-up and monitoring. The authors are acknowledged experts, and the book is a valuable desk reference for all who deal with inherited metabolic diseases. Chapter 73 is available open access under a Creative Commons Attribution 4.0 International License via link.springer.com

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