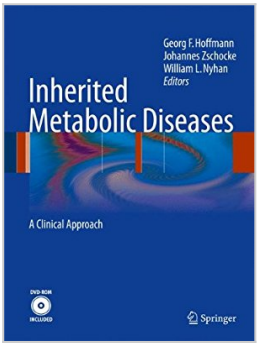

Inherited Metabolic Diseases: A Clinical Approach

From Springer

Inherited Metabolic Diseases: A Clinical Approach From Springer

The ? eld of inherited metabolic diseases has changed from a limited group of rare, untreatable, often fatal disorders to an important cause of acutely life-threatening but increasingly treatable illness. Unchanged is the orphan nature of these disorders with mostly relatively nonspeci? c initial clinical manifestations. The patient does not come to the physician with the diagnosis; the patient comes with a history, symptoms, and signs. This book starts with those and proceeds locally through algorithms from questions to answers. Special emphasis is placed on acutely presenting disorders and emergency situations. The rationale of the approaches presented in this book are based on extensive, collective clinical experience. To utilize as broad an experience as possible, its concept has been extended from a pocket-size book written jointly by ? ve colleagues to a textbook combining the experience of over 20 expert metabolic physicians. It is now imbedded in the environment of Springer Pediatric Metabolic Medicine in addition to the disease-based approach in Inborn Metabolic Diseases edited by John Fernandes and colleagues as well the series edited by Nenad Blau and colleagues on speci? c biochemical diagnostics, laboratory methods, and treatment. A system and symptom-based approach to inherited metabolic diseases should help colleagues from different specialties to diagnose their patients and to come to an optimal program of therapy. For metabolic and genetic specialists, this book is designed as a quick reference for what may be (even for the specialist) infrequently encountered presentations. Heidelberg, Germany Georg F.

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Editorial Review

Review

From the reviews:

“The authors have written a reader friendly and instructive textbook which is divided into 5 parts. … Highly illustrated with many tables. … An updated, very useful text.” (Pediatric Endocrinology Reviews, Vol. 7 (3), March-April, 2010)

“This is the eighth edition of an intense but easy to read textbook of genetics with detailed explanations of the concepts and logic behind the science of genes and genomes. … Designed for undergraduate students, this book provides a concise and logical base for individuals who may pursue careers in genetics. … The authors are well recognized authorities in human genetics. … Congratulations to the authors for this unique textbook. The art of teaching genetics is well served by their contribution.” (Luis F. Escobar, Doody’s Review Service, February, 2012)

From the Back Cover

This book focuses on clinical presentations that may be caused by inherited metabolic diseases. Its symptom- and system-based approach will help clinicians with and without detailed knowledge of human biochemistry in all specialties to reach a correct diagnosis and institute the optimal treatment program. The book summarizes the central elements of inherited metabolic diseases and describes clearly how to carry out an efficient yet complete diagnostic work-up, thereby guiding the clinician from the presenting symptoms and signs through to effective initial management. After an introduction to the different disorders, the book explains when to consider an inborn metabolic error and which initial tests to order. Core aspects such as structured communication, guidelines, transition, pregnancy, maternal care and how to respond to various medical emergencies are covered. Therapeutic concepts such as dietary treatment are delineated and practical advice provided on the quite different treatment approaches required for individual diseases. An extensive section structured according to organ systems outlines the correct approach in the context of specific symptoms and signs. The value of each of the potential investigations is explained, with precise advice on the interpretation of results. The inclusion of algorithms, tables, lists, and charts facilitates rapid decision making and information retrieval, and the appendices include a helpful guide to differential diagnosis based on clinical and biochemical phenotypes. This new updated edition of Inherited Metabolic Diseases will be an invaluable aid for the busy clinician and an excellent quick reference for metabolic and genetic specialists.

About the Author

Georg F. Hoffmann, Dr.med.habil, MD, is Professor and Chairman of the University Childrens Hospital Heidelberg, Head of the Metabolic Center including the Newborn Screening Laboratory as well as Head of the Center for Rare Diseases, Medical Center University of Heidelberg Germany. He has been working in the field of diagnosis and treatment of patients with inherited metabolic diseases for over 30 years with the main emphasis on newborn screening as well as clinical and laboratory research on neurometabolic and intoxication type disorders (neurotransmitter defects, aminoacidopathies, organoacidopathies, and urea cycle disorders).

Johannes Zschocke, Dr.med.habil, PhD is Professor and Chair of Human Genetics at the Medical University Innsbruck, Austria, and is Director of the Division for Human Genetics and the Center for Medical Genetics Innsbruck. He has longstanding clinical and research experience in inherited metabolic diseases, with special expertise in genetic diagnosis and genotype-phenotype correlations.

William L. Nyhan, MD, PhD is Distinguished Professor of Pediatrics and Director of the Biochemical Genetics Laboratory at the University of California San Diego. He is the author of 655 publications, including the Atlas of Inherited Metabolic Diseases and is a Board Member of Lesch-Nyhan Syndrome Children's Research Foundation, 1995 to present.

Users Review

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Willie Collier:

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Margaret Soto:

With this era which is the greater man or woman or who has ability in doing something more are more precious than other. Do you want to become one of it? It is just simple strategy to have that. What you are related is just spending your time not very much but quite enough to have a look at some books. Among the books in the top listing in your reading list is actually Inherited Metabolic Diseases: A Clinical Approach. This book that is qualified as The Hungry Inclines can get you closer in growing to be precious person. By looking right up and review this publication you can get many advantages.

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