



Inherited Metabolic Diseases: A Clinical Approach

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The field 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 nonspecific 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 five 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 specific 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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