

BOOK REVIEW

# A practical guide for your daily activity

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**Atlas of Metabolic Diseases – Second Edition**

Edited by William L. Nyhan, Bruce A. Barshop and Pinar T. Ozand. Hodder Arnold – a member of the Hodder Headline Group, distributed in the United States of America by Oxford University Press Inc., 2005. pp. 788. £150.00/US\$260 (approx.)/€220 (approx.) (Hardback).  
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It was a great pleasure reviewing this book due to the practical, comprehensive and attractive way to present such a complex material. This second edition represents an updated and completed version of the original variant of 'Atlas of Metabolic Disorders' published by Nyhan and Ozand in 1999 (Figure 1). As this field is rapidly moving, many chapters have been added or completed in the new volume.

The material is structured in 14 parts covering organic acidemias, disorders of amino-acid metabolism, urea cycle and fatty acid oxidation, lactic acidemias and mitochondrial disease, disorders of carbohydrate metabolism, peroxisomal disorders, disorders of purine metabolism, transport and mineral metabolism, mucopolysaccharidoses, mucopolipidoses, disorders of cholesterol and neutral lipid metabolism and lipid storage disorders. Although the subject seems difficult, the authors have achieved an outstanding presentation of 112 metabolic disorders in a very readable manner.

In the beginning of every part, the authors have inserted a short illustration of the biochemical pathway involved, making the material easy to understand. Each chapter is divided into 'Introduction', 'Clinical abnormalities', 'Genetics and pathogenesis' and 'Treatment', but probably the most useful for a quick reference is the introductory box that highlights the main features, which the metabolic disease may present itself with.

The text combines molecular biology, genetics, metabolic pathways and clinical practice, but the focus on the clinician is more than obvious. Algorithms and the rich selection of illustrations (clinical

photographs, as well as radiological and histological investigations) make the material practical and instructive. When I first examined the 'Atlas of Metabolic Diseases', the illustrations made me think of my own patients and I was eager to find out more. I have also appreciated the treatment sections – methods, products, consequences as well as the management directions that are presented briefly, for the benefit of the clinician who is trying to find out 'everything' on a specific disorder.

Another particularly useful aspect for quick reference is the appendix that includes the 'Index of disorders' and also the 'Index of signs and symptoms'. These are completed with a very practical 'Differential diagnosis of clinical phenotypes' section that lists for every prominent clinical feature/biochemical abnormality the metabolic disorders involved. Surprisingly, this part doesn't mention the page for every disorder, making the search more difficult for an inexperienced reader.

The bibliography was thoroughly selected for every chapter – historical articles, crucial for the description of each entity, as well as very recent ones are listed as they are used in the text. Maybe an alphabetical listing would be more useful for a quick reference.

In conclusion, this is an excellent book that should be on all pediatric wards, neonatal units and Medical Genetics Centers, outpatient departments as well as medical school libraries. The Atlas could be read from cover to cover (as the authors suggest for those preparing for examinations), but in my opinion this book should be leafed through when faced with a diagnostic dilemma. Once a diagnostic is established, the volume provides excellent information regarding clinical picture, investigations, genetics and management ■

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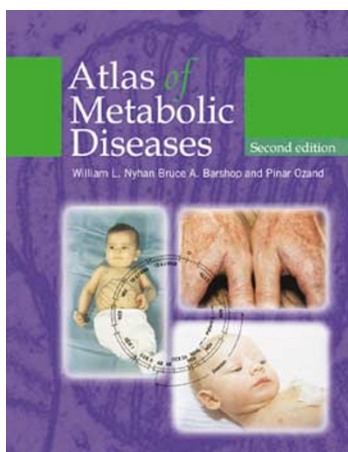


Figure 1