

Inborn metabolic diseases : diagnosis and treatment / Jean-Marie Saudubray, Georges van den Berghe, John H. Walter, (editors)

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Abstrak

For those involved in the identification and management of patients with inborn errors of metabolism, this book is now recognised as the standard textbook in this interdisciplinary field. It has proved to be indispensable for professionals in specialities ranging from pediatrics, neonatology, pathological biochemistry and genetics to neurology, internal medicine, nursing, dietetics and psychology. This 5th edition has been extensively revised and updated.

What's new

- Additional chapter focusing on inborn errors affecting adults, particularly the late neurological presentations
- Numerous updates on diagnostic procedures and treatment
- Newly discovered disorders.

As with previous editions, the book opens with a section presenting the clinical approach to inborn metabolic diseases for those cases in which a diagnosis is being sought. This now includes a chapter on the clinical presentation of metabolic diseases in the older age range, using an analogous structure. If a particular diagnosis is already suspected, the reader may refer to the following sections of the book containing general chapters on diagnostic procedures and treatment, and on specific groups of disorders.