



# Inborn Metabolic Diseases: Diagnosis and Treatment

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# Inborn Metabolic Diseases: Diagnosis and Treatment

## **Inborn Metabolic Diseases: Diagnosis and Treatment**

The editors of *Inborn Metabolic Diseases: Diagnosis and Treatment* are joined by coauthors from a world-class cadre to produce a book about Mendelian disorders of metabolism. The emphasis is on practical matters: diagnosis of the clinical and chemical manifestations of this type of disease in both its general and its particular forms, and treatment of the patient with the disease. The authors are concerned primarily with the consequences (manifestations of the associated disease) of deranged metabolic homeostasis (pathogenesis) that originate in mutation(s) of major genes (causes of the diseases). This book ought to be the ideal companion to *Metabolic Basis of Inherited Disease* (6th edition, 1989) in which the molecular causes and pathogenesis of inborn errors of metabolism are emphasized. Both books reveal the attitudes and methods that characterize this branch of medicine and of medical genetics in particular: both also - whether they intended to or not - disclose a gap between theory and practice in the discipline.

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