

Inborn Metabolic Diseases: Diagnosis and Treatment



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The editors of Inborn Metabolic Diseases: Diagnosis and Treat ment are joined by coauthors from a worldclass cadre to pro duce 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 gen eral and its particular forms, and treatment of the patient with the disease. The authors are concerned primarily with the con sequences (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 ()/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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