Clinical Guide To Inherited Metabolic Diseases

This user-friendly clinical handbook provides an overview of how to go about recognizing and diagnosing inherited metabolic diseases.This user-friendly clinical handbook provides a clear and concise overview of how to go about recognizing and diagnosing inherited metabolic diseases. The reader is led through the diagnostic process from the identification of those features of an illness suggesting that it might be metabolic through the selection of appropriate laboratory investigation to a final diagnosis. The book is organized into chapters according to the most prominent presenting problem of patients with inherited metabolic diseases: neurologic, hepatic, cardiac, metabolic acidosis, dysmorphism, and acute catastrophic illness in the newborn. It also includes chapters on general principles, laboratory investigation, neonatal screening, and the principles of treatment. This new edition includes much greater depth on mitochondrial disease and congenital disorders of glycosylation. The chapters on neurological syndrome and newborn screening are greatly expanded, as are those on laboratory investigation and treatment, to take account of the very latest technological developments. EAN/ISBN : 9780511133244 Publisher(s): Cambridge University Press Format: ePub/PDF Author(s): Clarke, Joe T. R.

DOWNLOAD HERE