



**Pediatric Neurology Part III: Chapter 160.  
Diagnostic work-up in acute conditions of inborn  
errors of metabolism and storage diseases  
(Handbook of Clinical Neurology)**

*Valayannopoulos Vassili, Poll-The Bwee Tien*

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Inborn errors of metabolism may present with acute neurological symptoms at any age. However, especially in neonates and infants, these conditions may be acute and if untreated may lead to permanent cerebral lesions or to death. Knowledge of the main signs and symptoms of these conditions may be lifesaving, especially for conditions that are treatable. From the pathophysiological perspective, errors of metabolism can be divided into disorders causing “intoxication,” disorders impairing energy production, and disorders involving complex molecules. From the clinical perspective, errors of metabolism may present with acute symptoms in the neonatal period and early infancy; late-onset acute and recurrent attacks; chronic and progressive symptoms. Nonspecific readily available biochemical markers may suggest the underlying condition but in most cases the choice of appropriate biochemical and molecular tests is required to establish the diagnosis. Progress in the treatment of inborn errors of metabolism has been slower than progress in diagnostic methods and in understanding of the pathophysiology of these disorders. Nevertheless, outcomes are improving with the use of dialysis and drugs to promote the removal of toxic metabolites and measures to keep catabolism to a minimum. Early intervention is crucial when neurological sequelae could be avoided, which requires constant vigilance and routine measurement of diagnostic biochemical markers in suspected cases.

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