Pediatric Neurology Part I: Chapter 56. Epilepsy in inborn errors of metabolism (Handbook of Clinical Neurology)

Nadia Bahi-buisson, Olivier Dulac

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Epilepsies associated with inborn errors of metabolism (IEM) represent a major challenge. Seizures rarely dominate the clinical presentation, which is more frequently associated with other neurological symptoms, such as hypotonia and/or cognitive disturbances. Although epilepsy in IEM can be classified in various ways according to pathogenesis, age of onset, or electroclinical presentation, the most pragmatic approach is determined by whether they are accessible to specific treatment or not. The main potentially treatable causes comprise vitamin B6 (pyridoxine deficiency), biotine, and GLUT1 deficiency (GLUT1DS) syndromes. Folinic acid-dependent seizures are allelic with pyridoxine dependency. Incompletely treatable IEMs include pyridoxal phosphate, serine, and creatine deficiencies. The main IEMs that present with epilepsy but offer no specific treatment are nonketotic hyperglycinemia, mitochondrial disorders, sulfite oxidase deficiency, ceroid-lipofuscinosis, Menkes disease, and peroxisomal disorders.

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