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Childhood encephalopathy due to thiamine pyrophosphokinase deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Childhood encephalopathy due to thiamine pyrophosphokinase deficiency. ORPHA:293955*

Childhood encephalopathy due to thiamine pyrophosphokinase deficiency is a rare inborn error of metabolism disorder characterized by early-onset, acute, encephalopathic episodes (frequently triggered by viral infections), associated with lactic acidosis and alpha-ketoglutaric aciduria, which typically manifest with variable degrees of ataxia, generalized developmental regression (which deteriorates with each episode) and dystonia. Other manifestations include spasticity, seizures, truncal hypotonia, limb hypertonia, brisk tendon reflexes and reversible coma.