

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Kod ORPHA

293955

Kod OMIM

614458

Kod ICD10

G96.8

Kod ICD11

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*Źródło

orphanet