Gamma-aminobutyric acid transaminase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Gamma-aminobutyric acid transaminase deficiency. ORPHA:2066

Gamma-aminobutyric acid transaminase (GABA-T) deficiency is an extremely rare disorder of GABA metabolism characterized by a severe neonatal-infantile epileptic encephalopathy (manifesting with symptoms such as seizures, hypotonia, hyperreflexia and developmental delay) and growth acceleration.