Pterin-4 alpha-carbinolamine dehydratase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pterin-4 alpha-carbinolamine dehydratase deficiency. ORPHA:1578

Dehydratase deficiency or pterin-4 alpha-carbinolamine dehydratase (PCD) is considered a transient and benign form of hyperphenylalaninemia due to tetrahydrobiopterin deficiency (see this term), characterized by muscular hypotonia, irritability (detected by EEG), slow acquisition of psychomotor skills, age-dependent movement disorders, including dystonia and an accompanying excretion of 7-substituted pterins. Neurological developement is normal with dietary control of blood phenyalanine. PCD is inherited in an autosomal recessive manner.