Dihydropteridine reductase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Dihydropteridine reductase deficiency. ORPHA:226

Dihydropteridine reductase (DHPR) deficiency is a severe form of hyperphenylalaninemia (HPA) due to impaired regeneration of tetrahydrobiopterin (BH4) (see this term), leading to decreased levels of neurotransmitters (dopamine, serotonin) and folate in cerebrospinal fluid, and causing neurological symptoms such as psychomotor delay, hypotonia, seizures, abnormal movements, hypersalivation, and swallowing difficulties.