

Opis choroby *

Definicja

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.

Dane

Klasyfikacja	Synonimy
Podtyp kliniczny	Hyperphenylalaninemia due to dihydropteridine reductase deficiency Fenyloketonuria typu 2 Hiperfenyloalaninemia spowodowana niedoborem reduktazy dihydropterydyny PKU typu 2 PKU type 2 Phenylketonuria type 2

Kod ORPHA 226	Kod OMIM 261630	Kod ICD10 E70.1
Kod ICD11 5C59.01		

*Źródło

orphanet