Opis choroby *

Definicja

An amino acid disorder with neonatal onset that is clinically characterized by the classic manifestations of phenylketonuria (PKA) and that later on is clinically differentiated by neurologic symptoms such as microcephaly, intellectual disability, central hypotonia, delayed motor development, peripheral spasticity and seizures, that develop and persist despite an established metabolic control of plasma phenylalanine.

Dane

Klasyfikacja
Choroba

Synonimy
Hyperphenylalaninemia due to BH4 deficiency
Hiperfenylalaninemia nie związana z fenyloketonurią
Hiperfenyloalaninemia z powodu niedoboru BH4
Hiperfenyloalaninemia z powodu niedoboru tetrahydrobiopteryny
Non-phenylketonuric hyperphenylalaninemia

Kod ORPHA
238583

Kod OMIM
261640

Kod ICD10
E70.1

Kod ICD11
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*Źródło

orphanet