

Opis choroby *

Definicja

A rare inborn error of metabolism characterized by increased serum phenylalanine, associated with variable neurological symptoms ranging from mild autistic features or hyperactivity to severe intellectual disability, dystonia, and parkinsonism. Laboratory analyses show normal tetrahydrobiopterin (BH4) metabolism and low levels of the CSF monoamine neurotransmitter metabolites homovanillic acid and 5-hydroxyindoleacetic acid.

Dane

Klasyfikacja

Choroba	Synonimy Non-phenylketonuric non-BH4-deficiency hyperphenylalaninemia Hiperfenylalaninemia inna niż fenyloketonuria i niedobór BH4
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Kod ORPHA

508523

Kod OMIM

617384

Kod ICD10

E70.1

Kod ICD11

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*Źródło

orphanet