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

Kod ORPHA
508523

Kod OMIM
617384

Kod ICD10
E70.1

Kod ICD11
-

---

*Źródło

orphanet