Opis choroby *

Definicja

A rare disorder of phenylalanine (Phe) metabolism, an inborn error of amino acid metabolism, characterized by the development of microcephaly, growth retardation, congenital heart disease, facial dysmorphism and intellectual disability in non-phenylketonuric offspring of mothers with excess blood Phe concentrations.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Hyperphenylalaninemic embryopathy
Embriopatia fenyloketonowa
Embriopatia hiperfenyloalaninowa
Hiperfenyloalaninemia matczyna
PKU matczyna
Maternal PKU
Maternal hyperphenylalaninemia
Phenylketonuric embryopathy

Kod ORPHA
2209

Kod OMIM
261600

Kod ICD10
E70.1

Kod ICD11
5C50.02

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*Źródło

orphanet