

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

[*Źródło](#)

orphanet