

Opis choroby *

Definicja

A rare inborn error of metabolism characterized by congenital hypertryptophanemia and hyperserotonemia. Patients are typically asymptomatic, although developmental delay, intellectual disability, and behavioral abnormalities, among others, have been reported in association.

Dane

Klasyfikacja

Choroba

Kod ORPHA

2224

Kod OMIM

600627

Kod ICD10

E70.8

Kod ICD11

5C50.3

*Źródło

orphanet