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

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*Źródło

orphanet