Opis choroby *

Definicja

A rare inborn error of metabolism characterized by infantile onset of global developmental delay, severe intellectual disability, seizures, and movement disorder (including tremor, hyperkinesia, and myoclonus), associated with excessive excretion of hydroxylysine in urine. There have been no further descriptions in the literature since 1970.

Dane

Klasyfikacja
Choroba

Kod ORPHA
79156

Kod OMIM
236900

Kod ICD10
E72.3

Kod ICD11
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*Źródło

orphanet