Opis choroby *

Definicja

A rare classic organic aciduria characterized by tissue accumulation and elevation of urinary excretion of 3-hydroxyisobutyric acid. The clinical phenotype ranges from recurrent mild episodes of vomiting with normal cognitive development, to massive acidosis, seizures, and failure to thrive with profound intellectual disability and early death. Dysmorphic craniofacial features (such as microcephaly, triangular face, short, sloping forehead, long, prominent philtrum, and micrognathia) and variable cerebral anomalies have also been described.

Dane

Klasyfikacja
Choroba

Kod ORPHA
939

Kod OMIM
236795

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet