

# Kwasica 3-hydroksyizomastowa

## Kod Orpha: 939 Kod OMIM: 236795

### 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

### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.