

Opis choroby *

Definicja

Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency is characterised by delayed motor development, hypotonia and progressive neurodegeneration. To date, it has been described in four boys. The syndrome is caused by mutations affecting the two alleles of the <i>HIBCH</i> gene, encoding 3-hydroxyisobutyryl-CoA hydrolase. The mode of transmission has not yet been established.

Dane

Klasyfikacja

Choroba

Synonimy

HIBCH deficiency
Acyduria metakrylowa
Defekt metabolizmu waliny
Niedobór HIBCH
Methacrylic aciduria
Valine metabolic defect

Kod ORPHA

88639

Kod OMIM

250620

Kod ICD10

E71.1

Kod ICD11

5C50.E0

*Źródło

orphanet