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 HIBCH 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

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*Źródło

orphanet