

Opis choroby *

Definicja

HSD10 disease is a rare, life-threatening neurometabolic disease characterized by a progressive neurodegenerative course, epilepsy, retinopathy and progressive cardiomyopathy.

Dane

Klasyfikacja

Choroba

Synonimy

2-methyl-3-hydroxybutyric aciduria
Acyduria 2-metylo-3-hydroksymasłowa
Niedobór dehydrogenazy 2-metylo-3-
hydroksymasłowego-CoA
Niedobór HSD10
Niedobór MHBD
2-methyl-3-hydroxybutyryl-CoA dehydrogenase
deficiency
HSD10 deficiency
MHBD deficiency

Kod ORPHA

391417

Kod OMIM

300438

Kod ICD10

E72.8

Kod ICD11

5C52.01

[*Źródło](#)

orphanet