

## 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

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#### [\\*Źródło](#)

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