

## Opis choroby \*

### Definicja

Ethylmalonic acid encephalopathy (EE) is defined by elevated excretion of ethylmalonic acid (EMA) with recurrent petechiae, orthostatic acrocyanosis and chronic diarrhoea associated with neurodevelopmental delay, psychomotor regression and hypotonia with brain magnetic resonance imaging (MRI) abnormalities.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

51188

#### Kod OMIM

602473

#### Kod ICD10

E88.8

#### Kod ICD11

5C50.E1

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### \*Źródło

orphanet