

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

*Źródło

orphanet