

Opis choroby *

Definicja

Homocystinuria due to methylene tetrahydrofolate reductase (MTHFR) deficiency is a metabolic disorder characterised by neurological manifestations.

Dane

Klasyfikacja

Choroba

Synonimy

MTHFR deficiency

Niedobór MTHFR

Niedobór reduktazy metylenotetrahydrofolianu

Methylene tetrahydrofolate reductase deficiency

Kod ORPHA

395

Kod OMIM

236250

Kod ICD10

E72.1

Kod ICD11

5C63.1

*Źródło

orphanet