

Opis choroby *

Definicja

Classical homocystinuria due to cystathionine beta-synthase (CbS) deficiency is characterized by the multiple involvement of the eye, skeleton, central nervous system, and vascular system.

Dane

Klasyfikacja

Choroba

Synonimy

Cystathionine beta-synthase deficiency
Homocystynuria spowodowana niedoborem
beta-syntazy cystationu
Niedobór beta-syntazy cystationu
Homocystinuria due to cystathionine beta-
synthase deficiency

Kod ORPHA

394

Kod OMIM

236200

Kod ICD10

E72.1

Kod ICD11

5C50.B

*Źródło

orphanet