

## 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

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#### \*Źródło

orphanet