

Opis choroby *

Definicja

Desmosterolosis is a very rare sterol biosynthesis disorder characterized by multiple congenital anomalies, failure to thrive, and intellectual disability, with elevated levels of desmosterol.

Dane

Klasyfikacja

Choroba

Kod ORPHA

35107

Kod OMIM

602398

Kod ICD10

Q87.8

Kod ICD11

5C52.10

*Źródło

orphanet