

Opis choroby *

Definicja

Lathosterolosis is an extremely rare inborn error of sterol biosynthesis characterized by facial dysmorphism, congenital anomalies (including limb and kidney anomalies), failure to thrive, developmental delay and liver disease.

Dane

Klasyfikacja

Choroba

Synonimy

Sterol C5-desaturase deficiency

Niedobór C5-desaturazy sterolowej

Kod ORPHA

46059

Kod OMIM

607330

Kod ICD10

Q87.8

Kod ICD11

5C52.10

*Źródło

orphanet