

Opis choroby *

Definicja

A rare developmental defect during embryogenesis characterized by microcephaly, characteristic facial features, hypotonia, non-progressive intellectual deficit, myopia and retinal dystrophy, neutropenia and truncal obesity.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

193

Kod OMIM

216550

Kod ICD10

Q87.8

Kod ICD11

LD90.Y

*Źródło

orphanet