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

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*Źródło

orphanet