

Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by severe intellectual disability, distinctive craniofacial features and variable multiple congenital anomalies including ocular, brain, urogenital and skeletal abnormalities.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

2707

Kod OMIM

244450

Kod ICD10

Q87.0

Kod ICD11

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*Źródło

orphanet