

Opis choroby *

Definicja

Tetrasomy 18p is a very rare structural chromosomal anomaly affecting multiple body systems and characterized clinically by craniofacial abnormalities, delayed development, cognitive impairment, changes in muscle tone, distinctive facial features, and rarely renal malformations.

Dane

Klasyfikacja

Zespół wad wrodzonych Isochromosome 18p
Isochromosom 18p

Kod ORPHA

3307

Kod OMIM

614290

Kod ICD10

Q99.8

Kod ICD11

LD7Y

*Źródło

orphanet