

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	Synonimy
Zespół wad wrodzonych	Isochromosome 18p
	Izochromosom 18p

Kod ORPHA
3307

Kod OMIM
614290

Kod ICD10
Q99.8

Kod ICD11
LD7Y

*Źródło

orphanet