Opis choroby *

Definicja

Tetrasomy 5p is a rare chromosomal anomaly syndrome with variable phenotype principally characterized by developmental delay, growth retardation/short stature, hypotonia, seizures, ventriculomegaly, hand and foot anomalies (e.g. clinodactyly, overlapping toes) and mosaic pigmentary skin changes. Patients may also present minor dysmorphic craniofacial features (incl. macrocephaly, upslanting palpebral fissures, hypertelorism, abnormal auricles, anteverted nasal tip, midface hypoplasia).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Isochromosome 5p
Izochromosom 5p

Kod ORPHA
3309

Kod OMIM
-

Kod ICD10
Q99.8

Kod ICD11
-

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*Źródło

orphanet