Opis choroby *

Definicja

Trisomy 4p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 4, with a highly variable phenotype, typically characterized by pre- and postnatal growth delay, psychomotor developmental delay and craniofacial dysmorphism (microcephaly, prominent glabelle, hypertelorism, enlarged ears with abnormal helix and antihelix, bulbous nose with flat or depressed nasal bridge, long philtrum, retrognathia with pointed chin). Additional features include skeletal (rocker bottom feet, arachnodactyly, camptodactyly) and renal malformations, cardiac defects, ocular abnormalities and abnormal genitalia in males.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Duplication 4p
Duplikacja 4p
Duplikacja krótkiego ramienia chromosomu 4
Trisomia krótkiego ramienia chromosomu 4
Duplication of the short arm of chromosome 4
Trisomy of the short arm of chromosome 4

Kod ORPHA
1738

Kod OMIM
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Kod ICD10
Q92.2

Kod ICD11
LD41.31

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*Źródło

orphanet