Opis choroby *

Definicja

Distal trisomy 3p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 3, with highly variable phenotype principally characterized by craniofacial dysmorphism (incl. brachy-/microcephaly, square facies, frontal bossing, bitemporal indentation, hypertelorism/telecanthus, low-set and/or dysmorphic ears, short nose with broad, flat nasal bridge, prominent cheeks and philtrum, downturned corners of mouth, micrognathia/retrognathia, short neck) associated with psychomotor delay, moderate to severe intellectual disability, cardiac (e.g. patent ductus arteriosus) and urogenital (e.g. renal hypoplasia, hypogenitalism) abnormalities, as well as seizures and presence of whorls on fingers.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 3p
Duplikacja dystalna 3p
Duplikacja telomerowa 3p
Trisomia 3pter
Telomeric duplication 3p
Trisomy 3pter

Kod ORPHA
96071

Kod OMIM
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Kod ICD10
Q92.3

Kod ICD11
LD41.21

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*Źródło

orphanet