Opis choroby *

Definicja

Distal trisomy 2p is a rare chromosomal anomaly syndrome, resulting from the partial duplication of the short arm of chromosome 2, with a highly variable phenotype principally characterized by pre- and post-natal growth failure, global developmental delay, facial dysmorphism (incl. high forehead/frontal bossing, abnormal ear shape and/or position, hypertelorism/telecanthus, broad/depressed nasal bridge) and ocular anomalies (e.g. exophthalmos, retinal hypopigmentation, optic nerve and foveal hypoplasia). Other reported anomalies include generalized hypotonia, pectus excavatum, long fingers and toes, syndactyly, congenital heart (e.g. ventricular and atrial septal defects) and neural tube defects, seizures, pulmonary hypoplasia, diaphragmatic hernia and urogenital anomalies.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Distal duplication 2p
Duplikacja dystalna 2p
Duplikacja telomerowa 2p
Trisomia 2pter
Telomeric duplication 2p
Trisomy 2pter

Kod ORPHA
96070

Kod OMIM
-

Kod ICD10
Q92.3

Kod ICD11
LD41.11

---

*Źródło

orphanet