Opis choroby *

Definicja

Mosaic trisomy 10 is a rare chromosomal anomaly syndrome, with a highly variable phenotype, principally characterized by growth delay, craniofacial dysmorphism (incl. prominent forehead, hypertelorism, upslanting palpebral fissures, blepharophimosis, low-set malformed large ears, high arched palate, cleft lip/palate, retrognathia) and cardiac, renal and skeletal (e.g. radial ray defects, scoliosis) malformations, with death usually ocurring neonatally or in early infancy. Other reported features include central nervous system and ear anomalies, as well as facial clefts and anal atresia.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 10
Trisomy 10 mosaicism
Mosaic trisomy chromosome 10
Trisomy 10 mosaicism

Kod ORPHA
96063

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
LD40.Y

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*Źródło

orphanet