Opis choroby *

Definicja

Mosaic trisomy 15 is a rare chromosomal anomaly syndrome principally characterized by intrauterine growth restriction, congenital cardiac anomalies (incl. ventricular and atrial septal defects, patent ductus arteriosus) and craniofacial dysmorphism (incl. hypertelorism, downslanting palpebral fissures, wide nasal bridge). Patients also present brain (e.g. hypoplastic cerebellum, ventricular asymmetry), renal (e.g. small dysplastic kidneys), and/or genital (undescended testis, small penis, hypoplastic labia majora) anomalies. Digital and skin pigmentation abnormalities have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 15
Trisomy 15 mosaicism

Kod ORPHA
1706

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
-

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*Źródło

orphanet