Opis choroby *

Definicja

Mosaic trisomy 12 is a rare chromosomal anomaly syndrome, with a highly variable phenotype, principally characterized by developmental or growth delay, short stature, craniofacial dysmorphism (e.g. turricephaly, tall forehead, downslanting palpebral fissures, posteriorly rotated and low set ears, narrow palate), congenital heart defects (e.g. atrial septal defect, patent ductus arteriosus), hypotonia, and pigmentary dysplasia. Scoliosis, hearing loss, facial/body asymmetry, and intellectual disability have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 12
Trisomy 12 mosaicism

Kod ORPHA
1698

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
-

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*Źródło

orphanet