Opis choroby *

Definicja

Mosaic trisomy 17 is a rare chromosomal anomaly syndrome, with a highly variable clinical presentation, mostly characterized by growth delay, intellectual disability, body asymmetry with leg length differentiation, scoliosis, and congenital heart anomalies (e.g. ventricular septal defect). Prenatal ultrasound findings include intrauterine growth retardation, nuchal thickening brain anomalies (e.g. cerebellar hypoplasia), pleural effusion and single umbilical artery. Patients with no associated malformations have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 17
Trisomy 17 mosaicism

Kod ORPHA
1711

Kod OMIM
-

Kod ICD10
Q92.1

Kod ICD11
-

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*Źródło

orphanet