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 Synonimy

Zespół wad wrodzonych Mosaic trisomy chromosome 17

Trisomy 17 mosaicism

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 1711
 Q92.1

Kod ICD11

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*Źródło

orphanet