Opis choroby *

Definicja

Mosaic trisomy 16 is a rare chromosomal anomaly syndrome with a highly variable phenotype ranging from minor anomalies with normal development to intrauterine growth retardation, abnormal skin pigmentation, craniofacial and body asymmetry, cardiac (e.g. ventricular septal defect) and genital (e.g. hypospadias, cryptorchidism) anomalies, scoliosis and hearing loss to neonatal death. Additional features observed include skeletal malformations (e.g. clino/polydactyly, talipes), mild facial dysmorphism, and developmental delay.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Mosaic trisomy chromosome 16
Trisomy 16 mosaicism

Kod ORPHA
1708

Kod OMIM
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Kod ICD10
Q92.1

Kod ICD11
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*Źródło

orphanet