

Opis choroby *

Definicja

Mosaic trisomy 9 is a rare chromosomal anomaly syndrome, with a highly variable phenotype, principally characterized by intellectual disability, growth and developmental delay, facial dysmorphism (incl. microphthalmia, deep-set eyes, low-set, malformed ears, bulbous nose, high-arched palate, micrognathia) and congenital heart defects (e.g. ventricular septal defect), as well as urogenital (e.g. hypoplastic genitalia, cryptorchidism), skeletal (congenital joint dislocations or hyperflexion, scoliosis/kyphosis) and central nervous system anomalies (hydrocephalus, Dandy-Walker malformation). Pigmentary mosaic skin lesions along the lines of Blaschko are also frequently observed.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Mosaic trisomy chromosome 9 Trisomy 9 mosaicism Mosaic trisomy chromosome 9 Trisomy 9 mosaicism

Kod ORPHA 99776	Kod OMIM -	Kod ICD10 Q92.1
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Kod ICD11
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*Źródło

orphanet