

Opis choroby *

Definicja

Axial mesodermal dysplasia spectrum is a rare developmental defect during embryogenesis syndrome characterized by congenital manifestations of both oculo-auriculo-vertebral spectrum and caudal regression sequence. Phenotype is highly variable but patients typically present facial dysmorphism (incl. asymmetry, hypertelorism), auricular abnormalities (e.g. preauricular tags, microtia, absence of middle ear ossicles), skeletal malformations (hemivertebrae, hip dislocation, sacral agenesis/dysplasia, talipes equinovarus, flexion deformity of lower limbs), cardiac defects (dextrocardia, septal defects), renal and genitourinary anomalies (such as renal agenesis/dysplasia, abnormal external genitalia, cryptorchidia), as well as anal anomalies such as anal atresia and rectovesical fistula.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Blastogenesis defect
	Defekt blastogenezy
	Zespół Russella, Weavera i Bulla
	Russell-Weaver-Bull syndrome

Kod ORPHA	Kod OMIM	Kod ICD10
1834	-	Q87.8

Kod ICD11

-

*Źródło

orphanet