

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

Zespół wad wrodzonych

Synonimy

Blastogenesis defect

Defekt blastogenezy

Zespół Russella, Weavera i Bulla

Russell-Weaver-Bull syndrome

Kod ORPHA

1834

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet