Opis choroby *

Definicja

TARP syndrome is a rare developmental defect during embryogenesis syndrome characterized by Robin sequence (micrognathia, glossoptosis, and cleft palate), atrial septal defect, persistence of the left superior vena cava, and talipes equinovarus. The phenotype is variable, some patients present with further dysmorphic characteristics (e.g. hypertelorism, ear abnormalities) while others do not have any key findings. Additional features, such as syndactyly, polydactyly, or brain anomalies (e.g. cerebellar hypoplasia), have also been reported. The syndrome is almost invariably lethal with affected males either dying prenatally or living just a few months.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Pierre Robin sequence-congenital heart defect-talipes syndrome
Sekwencja Pierre'a i Robina - wrodzona wada serca - stopa końsko-szpotawa
Stopa końsko-szpotawa - wada przegrody międzyprzedsionkowej - sekwencja Robina - przetrwała żyła główna górna lewa
Zespół Pierre'a i Robina - wrodzona wada serca - stopa końsko-szpotawa
Pierre Robin syndrome-congenital heart defect-talipes syndrome
Talipes equinovarus-atrial septal defect-Robin sequence-persistence of the left superior vena cava syndrome

Kod ORPHA
2886

Kod OMIM
311900

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet