Opis choroby *

Definicja

A rare disorder characterized by mirror polydactyly, vertebral hypersegmentation and severe congenital limb deficiencies. Duodenal atresia and absent thymus were also reported. So far, it has been described in four unrelated infants identified through a congenital malformation screening program carried out in Spain. The prevalence was estimated at around 1 in 330,000. The etiology is unknown but it was suggested that the syndrome is caused by defective expression of a developmental control gene.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3004

Kod OMIM
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Kod ICD10
Q87.2

Kod ICD11
LD26.2

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*Źródło

orphanet