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Mirror polydactyly-vertebral segmentation-limbs defects syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Mirror polydactyly-vertebral segmentation-limbs defects syndrome. ORPHA:3004*

Mirror polydactyly-vertebral segmentation-limbs defects syndrome is characterised 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.