

Opis choroby *

Definicja

A rare syndrome of multiple congenital anomalies characterized by radial ray malformations, renal abnormalities (mild malrotation, ectopia, horseshoe kidney, renal hypoplasia, vesico-ureteral reflux, bladder diverticula), and ophthalmological abnormalities (mainly colobomas, but also microphthalmia, ptosis, and Duane anomaly). The phenotype overlaps with other *SALL4*-related disorders including Okihiro syndrome and Holt-Oram syndrome.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

959

Kod OMIM

607323

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet