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
-

---

*Źródło

orphanet