
DISEASE:
Acro-renal-ocular syndrome

NAME:	Acro-renal-ocular syndrome
DESCRIPTION:	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.
ORPHACODE:	959
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>SALL4</u>
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