

DISEASE:
Oculocerebrorenal syndrome of Lowe

NAME:	Oculocerebrorenal syndrome of Lowe
DESCRIPTION:	A rare multisystem disorder characterized by congenital cataracts, glaucoma, intellectual disabilities, seizures, postnatal growth retardation and renal tubular dysfunction with chronic renal failure.
ORPHACODE:	534
SYNONYMS:	Lowe disease Lowe oculo-cerebro-renal dystrophy Lowe oculo-cerebro-renal syndrome Lowe oculocerebrorenal dystrophy Lowe syndrome OCRL Phosphatidylinositol 4,5-biphosphate 5-phosphatase deficiency
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10
ANALYTE(S):	OCRL
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