DISEASE:
Renal coloboma syndrome

NAME:	Renal coloboma syndrome
DESCRIPTION:	A genetic condition characterized by optic nerve dysplasia and renal hypodysplasia.
ORPHACODE:	1475
SYNONYMS:	Coloboma of optic nerve with renal disease Papillo-renal syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PAX2
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Nephrotic syndrome, Focal Segmental Glomerulosclerosis (FSGS) , Alport syndrome and podocytopathy (gene panel)
- Renal or urinary tract malformation (CAKUT) (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- paired box 2

Related Gene Panels

- Cakut (congenital anomalies of the kidney and urinary tract-1) (69 genes) - IPG
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Nephrotic syndrome, FSGS, Alport syndrome (76 genes) - IPG