DISEASE:
Saldino-Mainzer syndrome

NAME:	Saldino-Mainzer syndrome
DESCRIPTION:	Saldino-Mainzer syndrome is characterised by the association of renal disease, retinal pigmentary dystrophy, cerebellar ataxia and skeletal dysplasia.
ORPHACODE:	140969
SYNONYMS:	Conorenal syndrome Renal dysplasia-retinal pigmentary dystrophy-cerebellar ataxia-skeletal dysplasia syndrome
XREF(S):	Orphanet OMIM OMIM MeSH ICD-10
ANALYTE(S):	IFT140 IFT172
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

Related Analytes

- intraflagellar transport 140
- intraflagellar transport 172

Related Gene Panels

- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL