GENETIC TEST:
Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)

FULL NAME:	Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Post-natal Diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS) Bi-directional Sanger Sequence analysis
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2022-10-07 / 2027-10-06

TURNAROUND TIME (MAXIMUM):	20 to 60 days
CREATED:	06 Aug 2019 - 11:46
CHANGED:	11 Dec 2023 - 13:00

RELATED CONTENT

Related Diseases

- ALG8-CDG
- Alagille syndrome due to a NOTCH2 point mutation
- Autosomal dominant polycystic kidney disease
- Autosomal dominant polycystic kidney disease type 1 with tuberous sclerosis
- Autosomal recessive polycystic kidney disease
- BOR syndrome
- Bardet-Biedl syndrome
- Birt-Hogg-Dubé syndrome
- Branchiootic syndrome
- COL4A1-related familial vascular leukoencephalopathy
- Caroli disease
- Cowden syndrome
- Familial idiopathic steroid-resistant nephrotic syndrome with focal segmental hyalinosis
- Familial juvenile hyperuricemic nephropathy type 1
- Genetic steroid-resistant nephrotic syndrome
- HANAC syndrome
- HNF1B-related autosomal dominant tubulointerstitial kidney disease
- Hereditary clear cell renal cell carcinoma
- Hereditary leiomyomatosis and renal cell cancer
- Hereditary papillary renal cell carcinoma
- Infantile nephronophthisis
- Isolated neonatal sclerosing cholangitis
- Isolated polycystic liver disease
- Isolated succinate-CoQ reductase deficiency
- Joubert syndrome
- Joubert syndrome with hepatic defect

- Joubert syndrome with ocular defect
- Joubert syndrome with oculorenal defect
- Joubert syndrome with renal defect
- Juvenile nephronophthisis
- Karyomegalic interstitial nephritis
- Late-onset nephronophthisis
- Meckel syndrome
- NPHP3-related Meckel-like syndrome
- Orofaciodigital syndrome type 1
- Orofaciodigital syndrome type 6
- PMM2-CDG
- Papillary renal cell carcinoma
- Renal coloboma syndrome
- Renal-hepatic-pancreatic dysplasia
- Saldino-Mainzer syndrome
- Senior-Boichis syndrome
- Senior-Loken syndrome
- UMOD-related autosomal dominant tubulointerstitial kidney disease
- Von Hippel-Lindau disease

Related Laboratories

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

Related Analytes

- ADAM metallopeptidase with thrombospondin type 1 motif 9
- Abelson helper integration site 1
- ALG5 dolichyl-phosphate beta-glucosyltransferase
- ALG8 alpha-1,3-glucosyltransferase

- ALG9 alpha-1,2-mannosyltransferase
- ankyrin repeat and sterile alpha motif domain containing 6
- ADP ribosylation factor like GTPase 13B
- ADP ribosylation factor like GTPase 3
- ADP ribosylation factor like GTPase 6
- armadillo repeat containing 9
- ataxin 10
- B9 domain containing 1
- B9 domain containing 2
- BRCA1 associated protein 1
- BBSome interacting protein 1
- Bardet-Biedl syndrome 1
- Bardet-Biedl syndrome 10
- Bardet-Biedl syndrome 12
- Bardet-Biedl syndrome 2
- Bardet-Biedl syndrome 4
- Bardet-Biedl syndrome 5
- Bardet-Biedl syndrome 7
- Bardet-Biedl syndrome 9
- C2 domain containing 3 centriole elongation regulator
- coiled-coil and C2 domain containing 2A
- coiled-coil domain containing 28B
- cell division cycle 73
- centrosomal protein 104
- centrosomal protein 120
- centrosomal protein 164
- centrosomal protein 290
- centrosomal protein 41
- centrosomal protein 83
- cilia and flagella associated protein 418
- collagen type IV alpha 1 chain
- collagen type IV alpha 3 chain
- collagen type IV alpha 4 chain

- collagen type IV alpha 5 chain
- ciliogenesis and planar polarity effector complex subunit 1
- centrosome and spindle pole associated protein 1
- cytochrome P450 family 24 subfamily A member 1
- doublecortin domain containing 2
- discs large MAGUK scaffold protein 5
- DnaJ heat shock protein family (Hsp40) member B11
- dynein cytoplasmic 2 heavy chain 1
- dynein 2 intermediate chain 1
- dynein 2 intermediate chain 2
- dynein cytoplasmic 2 light intermediate chain 1
- dynein light chain Tctex-type 2B
- DAZ interacting zinc finger protein 1 like
- EvC ciliary complex subunit 1
- EvC ciliary complex subunit 2
- EYA transcriptional coactivator and phosphatase 1
- family with sequence similarity 149 member B1
- FANCD2 and FANCI associated nuclease 1
- fumarate hydratase
- folliculin
- glucosidase II alpha subunit
- GATA binding protein 3
- glycine amidinotransferase
- GLIS family zinc finger 2
- HNF1 homeobox A
- HNF1 homeobox B
- HYLS1 centriolar and ciliogenesis associated
- intraflagellar transport 122
- intraflagellar transport 140
- intraflagellar transport 172
- intraflagellar transport 27
- intraflagellar transport 43
- intraflagellar transport 52

- intraflagellar transport 74
- intraflagellar transport 80
- intraflagellar transport 81
- inositol polyphosphate-5-phosphatase E
- inturned planar cell polarity protein
- inversin
- IQ motif containing B1
- jagged canonical Notch ligand 1
- katanin interacting protein
- KIAA0586
- KIAA0753
- kinesin family member 14
- kinesin family member 7
- LDL receptor related protein 5
- LDL receptor related protein 6
- leucine zipper transcription factor like 1
- mitogen-activated protein kinase binding protein 1
- MET proto-oncogene, receptor tyrosine kinase
- MKKS centrosomal shuttling protein
- MKS transition zone complex subunit 1
- NIMA related kinase 1
- NIMA related kinase 8
- notch receptor 2
- nephrocystin 1
- nephrocystin 3
- nephrocystin 4
- OFD1 centriole and centriolar satellite protein
- poly(A)-specific ribonuclease
- paired box 2
- phosphodiesterase 6D
- progesterone immunomodulatory binding factor 1
- polycystin 1, transient receptor potential channel interacting
- polycystin 2, transient receptor potential cation channel

- PKHD1 ciliary IPT domain containing fibrocystin/polyductin
- phosphomannomutase 2
- POC1 centriolar protein B
- protein kinase C substrate 80K-H
- phosphatase and tensin homolog
- renin
- ring finger protein 139
- RPGRIP1 like
- SHH signaling and ciliogenesis regulator SDCCAG8
- succinate dehydrogenase complex flavoprotein subunit A
- succinate dehydrogenase complex iron sulfur subunit B
- succinate dehydrogenase complex subunit C
- succinate dehydrogenase complex subunit D
- SEC61 translocon subunit alpha 1
- SEC61 translocon subunit beta
- SEC63 homolog, protein translocation regulator
- solute carrier family 41 member 1
- sterol regulatory element binding transcription factor 1
- SUFU negative regulator of hedgehog signaling
- tectonic family member 1
- tectonic family member 2
- tectonic family member 3
- transcription factor AP-2 alpha
- transmembrane protein 107
- transmembrane protein 138
- transmembrane protein 216
- transmembrane protein 218
- transmembrane protein 231
- transmembrane protein 237
- transmembrane protein 67
- TOG array regulator of axonemal microtubules 1
- TRAF3 interacting protein 1
- tripartite motif containing 32

- TSC complex subunit 1
- TSC complex subunit 2
- tetratricopeptide repeat domain 21B
- tetratricopeptide repeat domain 8
- thioredoxin domain containing 15
- uromodulin
- von Hippel-Lindau tumor suppressor
- WD repeat containing planar cell polarity effector
- WD repeat domain 19
- WD repeat domain 35
- X-prolyl aminopeptidase 3
- zinc finger protein 423

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
