

Full name:	Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
Type of panel:	<u>Custom panel</u>
Version number:	Version 7
Laboratory:	<u>Centre de Génétique-Institut de Pathologie et de Génétique (IPG)</u>
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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 Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ADAMTS9	100.00	1	NM_182920.2
AHI1	100.00	1	NM_001134831.2
ALG5	100.00	1	NM_013338.5
ALG8	100.00	1	NM_024079.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>ALG9</u>	100.00	1	NM_024740.2
<u>ANKS6</u>	100.00	1	NM_173551.5
<u>ARL13B</u>	100.00	1	NM_001174150.2
<u>ARL3</u>	100.00	1	NM_004311.4
<u>ARL6</u>	100.00	1	NM_001278293.3
<u>ARMC9</u>	100.00	1	NM_001352754.2
<u>ATXN10</u>	100.00	1	NM_013236.4
<u>B9D1</u>	100.00	1	NM_015681.6
<u>B9D2</u>	100.00	1	NM_030578.4
<u>BAP1</u>	100.00	1	NM_004656.4
<u>BBIP1</u>	100.00	1	NM_001195305.3
<u>BBS1</u>	100.00	1	NM_024649.5
<u>BBS10</u>	100.00	1	NM_024685.4
<u>BBS12</u>	100.00	1	NM_152618.3
<u>BBS2</u>	100.00	1	NM_031885.5
<u>BBS4</u>	100.00	1	NM_033028.5
<u>BBS5</u>	100.00	1	NM_152384.3
<u>BBS7</u>	100.00	1	NM_176824.3
<u>BBS9</u>	100.00	1	NM_198428.3
<u>C2CD3</u>	100.00	1	NM_001286577.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>CC2D2A</u>	100.00	1	NM_001378615.1
<u>CCDC28B</u>	100.00	1	NM_024296.5
<u>CDC73</u>	100.00	1	NM_024529.5
<u>CEP104</u>	100.00	1	NM_014704.4
<u>CEP120</u>	100.00	1	NM_001375405.1
<u>CEP164</u>	100.00	1	NM_014956.5
<u>CEP290</u>	100.00	1	NM_025114.4
<u>CEP41</u>	100.00	1	NM_018718.3
<u>CEP83</u>	100.00	1	NM_016122.3
<u>CFAP418</u>	100.00	1	NM_177965.4
<u>COL4A1</u>	100.00	1	NM_001845.6
<u>COL4A3</u>	100.00	1	NM_000091.5
<u>COL4A4</u>	100.00	1	NM_000092.5
<u>COL4A5</u>	100.00	1	NM_033380.3
<u>CPLANE1</u>	100.00	1	NM_001384732.1
<u>CSPP1</u>	100.00	1	NM_001382391.1
<u>CYP24A1</u>	100.00	1	NM_000782.5
<u>DCDC2</u>	100.00	1	NM_016356.5
<u>DLG5</u>	100.00	1	NM_004747.4
<u>DNAJB11</u>	100.00	1	NM_016306.6

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>DYNC2H1</u>	100.00	1	NM_001377.3
<u>DYNC2I1</u>	100.00	1	NM_018051.5
<u>DYNC2I2</u>	100.00	1	NM_052844.4
<u>DYNC2LI1</u>	100.00	1	NM_016008.4
<u>DYNLT2B</u>	100.00	1	NM_152773.5
<u>DZIP1L</u>	100.00	1	NM_173543.3
<u>EVC</u>	100.00	1	NM_153717.3
<u>EVC2</u>	100.00	1	NM_147127.5
<u>EYA1</u>	100.00	1	NM_000503.6
<u>FAM149B1</u>	100.00	1	NM_173348.2
<u>FAN1</u>	100.00	1	NM_014967.5
<u>FH</u>	100.00	1	NM_000143.4
<u>FLCN</u>	100.00	1	NM_144997.7
<u>GANAB</u>	100.00	1	NM_198334.3
<u>GATA3</u>	100.00	1	NM_001002295.2
<u>GATM</u>	100.00	1	NM_001482.3
<u>GLIS2</u>	100.00	1	NM_032575.3
<u>HNF1A</u>	100.00	1	NM_000545.8
<u>HNF1B</u>	100.00	1	NM_000458.4
<u>HYLS1</u>	100.00	1	NM_001134793.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>IFT122</u>	100.00	1	NM_052989.3
<u>IFT140</u>	100.00	1	NM_014714.4
<u>IFT172</u>	100.00	1	NM_015662.3
<u>IFT27</u>	100.00	1	NM_001177701.3
<u>IFT43</u>	100.00	1	NM_001102564.3
<u>IFT52</u>	100.00	1	NM_016004.5
<u>IFT74</u>	100.00	1	NM_025103.4
<u>IFT80</u>	100.00	1	NM_020800.3
<u>IFT81</u>	100.00	1	NM_014055.4
<u>INPP5E</u>	100.00	1	NM_019892.6
<u>INTU</u>	100.00	1	NM_015693.4
<u>INVS</u>	100.00	1	NM_014425.5
<u>IQCB1</u>	100.00	1	NM_001023570.4
<u>JAG1</u>	100.00	1	NM_000214.3
<u>KATNIP</u>	100.00	1	NM_015202.5
<u>KIAA0586</u>	100.00	1	NM_001329943.3
<u>KIAA0753</u>	100.00	1	NM_014804.3
<u>KIF14</u>	100.00	1	NM_014875.3
<u>KIF7</u>	100.00	1	NM_198525.3
<u>LRP5</u>	100.00	1	NM_002335.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>LRP6</u>	100.00	1	NM_002336.3
<u>LZTFL1</u>	100.00	1	NM_020347.4
<u>MAPKBP1</u>	100.00	1	NM_014994.3
<u>MET</u>	100.00	1	NM_000245.4
<u>MKKS</u>	100.00	1	NM_170784.3
<u>MKS1</u>	100.00	1	NM_017777.4
<u>NEK1</u>	100.00	1	NM_001199397.3
<u>NEK8</u>	100.00	1	NM_178170.3
<u>NOTCH2</u>	99.00	1	NM_024408.4
<u>NPHP1</u>	100.00	1	NM_001128178.3
<u>NPHP3</u>	100.00	1	NM_153240.5
<u>NPHP4</u>	100.00	1	NM_015102.5
<u>OFD1</u>	100.00	1	NM_003611.3
<u>PARN</u>	100.00	1	NM_002582.4
<u>PAX2</u>	100.00	1	NM_000278.5
<u>PDE6D</u>	100.00	1	NM_002601.4
<u>PIBF1</u>	100.00	1	NM_006346.4
<u>PKD1</u>	100.00	1	NM_001009944.3
<u>PKD2</u>	100.00	1	NM_000297.4
<u>PKHD1</u>	100.00	1	NM_138694.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>PMM2</u>	0.00	1	NM_000303.2 une seule position
<u>POC1B</u>	100.00	1	NM_172240.3
<u>PRKCSH</u>	100.00	1	NM_001289104.2
<u>PTEN</u>	100.00	1	NM_000314.8
<u>REN</u>	100.00	1	NM_000537.4
<u>RNF139</u>	100.00	1	NM_007218.4
<u>RPGRIP1L</u>	96.00	1	NM_015272.5
<u>SDCCAG8</u>	100.00	1	NM_006642.5
<u>SDHA</u>	100.00	1	NM_004168.4
<u>SDHB</u>	100.00	1	NM_003000.3
<u>SDHC</u>	100.00	1	NM_003001.5
<u>SDHD</u>	100.00	1	NM_003002.4
<u>SEC61A1</u>	100.00	1	NM_013336.4
<u>SEC61B</u>	100.00	1	NM_006808.3
<u>SEC63</u>	100.00	1	NM_007214.5
<u>SLC41A1</u>	100.00	1	NM_173854.6
<u>SREBF1</u>	100.00	1	NM_004176.5
<u>SUFU</u>	100.00	1	NM_016169.4
<u>TCTN1</u>	100.00	1	NM_001082538.3
<u>TCTN2</u>	100.00	1	NM_024809.5

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>TCTN3</u>	100.00	1	NM_015631.6
<u>TFAP2A</u>	100.00	1	NM_001372066.1
<u>TMEM107</u>	100.00	1	NM_183065.4
<u>TMEM138</u>	100.00	1	NM_016464.5
<u>TMEM216</u>	100.00	1	NM_001173990.3
<u>TMEM218</u>	100.00	1	NM_001258244.2
<u>TMEM231</u>	100.00	1	NM_001077418.3
<u>TMEM237</u>	100.00	1	NM_001044385.3
<u>TMEM67</u>	100.00	1	NM_153704.6
<u>TOGARAM1</u>	100.00	1	NM_001308120.2
<u>TRAF3IP1</u>	100.00	1	NM_015650.4
<u>TRIM32</u>	100.00	1	NM_012210.4
<u>TSC1</u>	100.00	1	NM_000368.5
<u>TSC2</u>	100.00	1	NM_000548.5
<u>TTC21B</u>	100.00	1	NM_024753.5
<u>TTC8</u>	100.00	1	NM_144596.4
<u>TXNDC15</u>	100.00	1	NM_024715.4
<u>UMOD</u>	100.00	1	NM_003361.4
<u>VHL</u>	100.00	1	NM_000551.4
<u>WDPCP</u>	100.00	1	NM_015910.7

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
<u>WDR19</u>	100.00	1	NM_025132.4
<u>WDR35</u>	100.00	1	NM_020779.4
<u>XPNPEP3</u>	100.00	1	NM_022098.4
<u>ZNF423</u>	100.00	1	NM_001379286.1