Export to PDF

Full name:	Tubulopathy/Nephrolithiasis (106 genes) - IPG
Type of panel:	Custom panel
Version number:	Version 7
Laboratory:	Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
Created:	04 Jul 2019 - 10:45
Changed:	11 Dec 2023 - 11:51

Related Diseases

- Adult hypophosphatasia
- Apparent mineralocorticoid excess
- Autosomal dominant distal renal tubular acidosis
- Autosomal dominant hypocalcemia
- Autosomal dominant hypophosphatemic rickets
- Autosomal dominant primary hypomagnesemia with hypocalciuria
- Autosomal recessive distal renal tubular acidosis
- Autosomal recessive hypophosphatemic rickets
- Autosomal recessive infantile hypercalcemia
- Autosomal recessive proximal renal tubular acidosis
- Bartter syndrome type 3
- Childhood-onset hypophosphatasia
- Cystinuria type A
- Distal renal tubular acidosis with anemia
- Dominant hypophosphatemia with nephrolithiasis or osteoporosis
- EAST syndrome
- Familial hypocalciuric hypercalcemia type 1
- Familial hypocalciuric hypercalcemia type 2
- Familial hypocalciuric hypercalcemia type 3
- Familial primary hypomagnesemia with normocalciuria and normocalcemia
- Generalized pseudohypoaldosteronism type 1

- Gitelman syndrome
- Hereditary hypophosphatemic rickets with hypercalciuria
- Hypohidrosis-electrolyte imbalance-lacrimal gland dysfunction-ichthyosis-xerostomia syndrome
- Hypotonia-cystinuria syndrome
- Idiopathic hypercalciuria
- Infantile hypophosphatasia
- Isolated autosomal dominant hypomagnesemia, Glaudemans type
- Nephrogenic diabetes insipidus
- Nephrogenic syndrome of inappropriate antidiuresis
- Primary Fanconi renotubular syndrome
- Primary hyperoxaluria type 1
- Primary hyperoxaluria type 2
- Primary hyperoxaluria type 3
- Primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement
- Primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement
- Primary hypomagnesemia with secondary hypocalcemia
- Pseudohypoaldosteronism type 2D
- Pseudohypoaldosteronism type 2E
- Pterin-4 alpha-carbinolamine dehydratase deficiency
- REN-related autosomal dominant tubulointerstitial kidney disease
- Renal pseudohypoaldosteronism type 1
- Renal tubular dysgenesis of genetic origin
- UMOD-related autosomal dominant tubulointerstitial kidney disease
- X-linked hypophosphatemia

Related Analytes

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ADCY10	100.00	1	NM_018417.6
AGT	100.00	1	NM_001384479.1
AGTR1	100.00	1	NM_000685.5
AGXT	100.00	1	NM_000030.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
ALDOB	100.00	1	NM_000035.4
ALPL	100.00	1	NM_000478.6
AP2S1	100.00	1	NM_004069.6
APRT	100.00	1	NM_000485.3
AQP2	100.00	1	NM_000486.6
ATP6V0A4	100.00	1	NM_020632.3
ATP6V1B1	100.00	1	NM_001692.4
ATP7B	100.00	1	NM_000053.4
AVPR2	100.00	1	NM_000054.7
BSND	100.00	1	NM_057176.3
CA2	100.00	1	NM_000067.3
CACNA1H	100.00	1	NM_021098.3
CASR	100.00	1	NM_000388.4
CDC73	100.00	1	NM_024529.5
CLCN2	100.00	1	NM_004366.6
CLCN5	100.00	1	NM_001127898.4
CLCNKA	100.00	1	NM_004070.4
CLCNKB	100.00	1	NM_000085.5
CLDN10	100.00	1	NM_006984.5
CLDN16	100.00	1	NM_006580.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
CLDN19	100.00	1	NM_148960.3
CNNM2	100.00	1	NM_017649.5
CTNS	100.00	1	NM_004937.3
CUL3	100.00	1	NM_003590.5
CYP11B1	100.00	1	NM_000497.4
CYP17A1	100.00	1	NM_000102.4
CYP24A1	100.00	1	NM_000782.5
DMP1	100.00	1	NM_004407.4
EGF	100.00	1	NM_001963.6
EGFR	100.00	1	NM_005228.5
EHHADH	100.00	1	NM_001966.4
ENPP1	100.00	1	NM_006208.3
FAH	100.00	1	NM_000137.4
FAM20A	100.00	1	NM_017565.4
FAN1	100.00	1	NM_014967.5
FANCA	100.00	1	no
FGF23	100.00	1	NM_020638.3
FOXI1	100.00	1	NM_012188.5
FXYD2	100.00	1	NM_001680.5
G6PC1	100.00	1	NM_000151.4

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
GALT	100.00	1	NM_000155.4
GATM	100.00	1	NM_001482.3
GNA11	100.00	1	NM_002067.5
GRHPR	100.00	1	NM_012203.2
HNF1A	100.00	1	NM_000545.8
HNF1B	100.00	1	NM_000458.4
HNF4A	100.00	1	NM_175914.5
HOGA1	100.00	1	NM_138413.4
HPRT1	100.00	1	NM_000194.3
HSD11B2	100.00	1	NM_000196.4
KCNJ1	100.00	1	NM_153766.3
KCNJ10	100.00	1	NM_002241.5
KCNJ16	100.00	1	NM_170741.4
KCNJ5	100.00	1	NM_000890.5
KL	100.00	1	NM_004795.4
KLHL3	100.00	1	NM_017415.3
LAGE3	100.00	1	NM_006014.5
LDHD	100.00	1	NM_194436.3
LRP2	100.00	1	NM_004525.3
MAGED2	100.00	1	NM_177433.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
MEN1	100.00	1	NM_001370259.2
MOCOS	100.00	1	NM_017947.4
NHERF1	100.00	1	NM_004252.5
NR3C2	100.00	1	NM_000901.5
OCRL	100.00	1	NM_000276.4
PCBD1	100.00	1	NM_000281.4
PHEX	100.00	1	NM_000444.6
PRPS1	100.00	1	NM_002764.4
REN	100.00	1	NM_000537.4
RRAGD	100.00	1	NM_021244.5
SCNN1A	100.00	1	NM_001038.6
SCNN1B	100.00	1	NM_000336.3
SCNN1G	100.00	1	NM_001039.4
SEC61A1	100.00	1	NM_013336.4
SLC12A1	100.00	1	NM_000338.3
SLC12A3	100.00	1	NM_001126108.2
SLC16A12	100.00	1	NM_213606.4
SLC22A12	100.00	1	NM_144585.4
SLC26A1	100.00	1	NM_022042.4
SLC2A2	100.00	1	NM_000340.2

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
SLC2A9	100.00	1	NM_020041.3
SLC34A1	100.00	1	NM_003052.5
SLC34A3	100.00	1	NM_001177316.2
SLC36A2	100.00	1	NM_181776.3
SLC3A1	100.00	1	NM_000341.4
SLC41A1	100.00	1	NM_173854.6
SLC4A1	100.00	1	NM_000342.4
SLC4A4	100.00	1	NM_001098484.3
SLC5A1	100.00	1	NM_000343.4
SLC5A2	100.00	1	NM_003041.4
SLC6A19	100.00	1	NM_001003841.3
SLC6A20	100.00	1	NM_020208.4
SLC7A9	100.00	1	NM_014270.5
TRPM6	100.00	1	NM_017662.5
UMOD	100.00	1	NM_003361.4
VDR	100.00	1	NM_000376.3
VIPAS39	100.00	1	NM_001193315.2
VPS33B	100.00	1	NM_018668.5
WDR72	100.00	1	NM_182758.4
WFS1	100.00	1	NM_006005.3

GENE	% OF CODING SEQUENCE SUFFICIENTLY COVERED TO DETECT HETEROZYGOUS MUTATIONS	COPY NUMBER VARIATION	COMMENTS
WNK1	100.00	1	NM_018979.4
WNK4	100.00	1	NM_032387.5
XDH	100.00	1	NM_000379.4