RENAL DISORDERS GENE PANEL

Gene symbol	Depth (reads)	Coverage (avg %)	OMIM disease	Description
ACTN4	94	80	603278	Glomerulosclerosis focal segmental 1
ADAMTS13	61	85	274150	Thrombotic thrombocytopenic purpura familial
AGTR1	147	100	145500	Hypertension essential
AGXT	88	95	259900	Hyperoxaluria primary type 1
AHI1	117	97	608629	Joubert syndrome-3
ALG8	91	96	608104	Congenital disorder of glycosylation type Ih
ALMS1	217	94	203800	Alstrom syndrome
AP2S1	93	95	600740	Hypocalciuric hypercalcemia familial type III
APRT	55	89	614723	Adenine phosphoribosyltransferase deficiency
AQP2	74	84	125800	Diabetes insipidus nephrogenic
ARL13B	113	94	612291	Joubert syndrome 8
ATP6V0A4	100	86	602722	Renal tubular acidosis distal autosomal recessive
ATP6V1B1	113	97	200	-
AVPR2	59	96	304800	Diabetes insipidus nephrogenic
B9D2	53	96	614175	Meckel syndrome 10
BBS10	128	100	209900	Bardet-Biedl syndrome 10
BBS12	163	100	209900	Bardet-Biedl syndrome 12
BBS2	130	91	209900	Bardet-Biedl syndrome 2
BBS4	106	87	209900	Bardet-Biedl syndrome 4
BBS9	118	95	209900	Bardet-Biedl syndrome 9
BCS1L	134	86	262000	Bjornstad syndrome
BICC1	115	88	200	-
BSND	109	89	602522	Bartter syndrome type 4a
CA2	157	87	259730	Osteopetrosis autosomal recessive 3 with renal tubular acidosis
CASR	125	96	239200	Hyperparathyroidism neonatal
CC2D2A	101	90	216360	COACH syndrome
CD2AP	114	96	607832	Glomerulosclerosis focal segmental 3
CEP164	89	87	614845	Nephronophthisis 15

CEP290	96	92	209900	Bardet-Biedl syndrome 14
CEP41	94	92	614464	Joubert syndrome 15
CFHR5	110	80	614809	Nephropathy due to CFHR5 deficiency
CLCN5	123	88	300009	Dent disease
CLCNKB	95	64	607364	Bartter syndrome type 3
CLDN16	146	92	248250	Hypomagnesemia 3 renal
CLDN19	70	89	248190	Hypomagnesemia 5 renal with ocular involvement
CNNM2	131	95	613882	Hypomagnesemia 6 renal
COL4A1	110	78	611773	Angiopathy hereditary with nephropathy aneurysms and muscle
COL4A3	82	83	104200	Alport syndrome autosomal dominant
COL4A4	100	83	203780	Alport syndrome autosomal recessive
COL4A5	64	87	301050	Alport syndrome
COQ2	79	93	607426	Coenzyme Q10 deficiency primary 1
COQ6	147	90	614650	Coenzyme Q10 deficiency primary 6
CTNS	116	94	219800	Cystinosis atypical nephropathic
CUBN	100	78	261100	Megaloblastic anemia-1 Finnish type
DGKE	109	91	615008	Nephrotic syndrome type 7
DMP1	117	99	241520	Hypophosphatemic rickets AR
EGF	124	89	611718	Hypomagnesemia 4 renal
EYA1	125	82	113650	Anterior segment anomalies with or without cataract
FAM58A	47	30	300707	STAR syndrome
FAN1	119	57	614817	Interstitial nephritis karyomegalic
FGF23	70	88	193100	Hypophosphatemic rickets autosomal dominant
FN1	108	80	601894	Glomerulopathy with fibronectin deposits 2
FRAS1	116	82	219000	Fraser syndrome
FREM1	121	87	608980	Bifid nose with or without anorectal and renal anomalies
FREM2	141	93	219000	Fraser syndrome
FXYD2	44	93	154020	Hypomagnesemia-2 renal
GATA3	133	94	146255	Hypoparathyroidism sensorineural deafness and renal dysplasia
GLA	82	80	301500	Fabry disease
GLB1	88	84	230500	GM1-gangliosidosis type I
GLIS2	95	94	611498	Nephronophthisis 7
GLIS3	104	88	610199	Diabetes mellitus neonatal with congenital hypothyroidism

GNA11	86	74	200	-
HNF1B	85	81	125853	Diabetes mellitus noninsulin-dependent
HPRT1	75	80	300323	HPRT-related gout
HSD11B2	105	89	218030	Apparent mineralocorticoid excess
IFT122	106	81	218330	Cranioectodermal dysplasia 1
IFT140	97	86	266920	Mainzer-Saldino syndrome
INF2	70	92	614455	Charcot-Marie-Tooth disease dominant intermediate E
INPP5E	67	95	213300	Joubert syndrome 1
INVS	128	90	602088	Nephronophthisis 2 infantile
IQCB1	94	73	609254	Senior-Loken syndrome 5
KAL1	87	81	308700	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)
KCNJ1	175	100	241200	Bartter syndrome type 2
KCNJ10	158	94	600791	Enlarged vestibular aqueduct digenic
KIF7	67	89	200990	Acrocallosal syndrome
LAMB2	108	94	614199	Nephrotic syndrome type 5 with or without ocular abnormalities
LCAT	98	91	136120	Fish-eye disease
LMX1B	92	88	161200	Nail-patella syndrome
LRP2	124	87	222448	Donnai-Barrow syndrome
LRP4	105	86	212780	Cenani-Lenz syndactyly syndrome
LYZ	119	93	105200	Amyloidosis renal
MAFB	74	100	166300	Multicentric carpotarsal osteolysis syndrome
MKKS	147	97	209900	Bardet-Biedl syndrome 6
MKS1	127	86	209900	Bardet-Biedl syndrome 13
MYH9	100	85	603622	Deafness autosomal dominant 17
MYO1E	108	86	614131	Glomerulosclerosis focal segmental 6
NEK1	110	96	263520	Short rib-polydactyly syndrome type IIA
NEK8	118	89	613824	Nephronophthisis 9
NOTCH2	113	86	610205	Alagille syndrome 2
NPHP1	117	93	609583	Joubert syndrome 4
NPHP3	112	95	267010	Meckel syndrome 7
NPHP4	94	82	606966	Nephronophthisis 4
NPHS1	89	88	256300	Nephrotic syndrome type 1
NPHS2	133	92	200	-

NR3C2	136	95	605115	Hypertension early-onset autosomal dominant with exacerbation in pregnancy
OCRL	107	90	300555	Dent disease 2
OFD1	62	54	300804	Joubert syndrome 10
PAX2	110	88	120330	Papillorenal syndrome
PDSS2	94	92	614652	Coenzyme Q10 deficiency primary 3
PHEX	104	94	307800	Hypophosphatemic rickets X-linked dominant
PKD1	59	91	173900	Polycystic kidney disease adult type I
PKD2	99	93	613095	Polycystic kidney disease 2
PKHD1	114	90	200	-
PLCE1	132	86	610725	Nephrotic syndrome type 3
PTH1R	86	95	200	-
PTPRO	110	90	614196	Nephrotic syndrome type 6
REN	98	88	613092	Hyperuricemic nephropathy familial juvenile 2
RPGRIP1L	100	92	216360	COACH syndrome
RRM2B	110	96	612075	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy)
SALL1	135	52	107480	Townes-Brocks branchiootorenal-like syndrome
SALL4	105	37	607323	Duane-radial ray syndrome
SARS2	75	92	613845	Hyperuricemia pulmonary hypertension renal failure and alkalosis
SCARB2	99	88	254900	Epilepsy progressive myoclonic 4 with or without renal failure
SCNN1A	89	93	613021	Bronchiectasis with or without elevated sweat chloride 2
SCNN1B	90	92	211400	Bronchiectasis with or without elevated sweat chloride 1
SCNN1G	135	97	613071	Bronchiectasis with or without elevated sweat chloride 3
SDCCAG8	96	88	613615	Senior-Loken syndrome 7
SIX1	80	94	608389	Brachiootic syndrome 3
SIX5	52	93	610896	Branchiootorenal syndrome 2
SLC12A1	144	92	601678	Bartter syndrome type 1
SLC12A3	88	95	263800	Gitelman syndrome
SLC16A12	132	93	612018	Cataract juvenile with microcornea and glucosuria
SLC22A12	100	84	220150	Hypouricemia renal
SLC26A3	131	87	214700	Chloride diarrhea congenital Finnish type
SLC2A2	139	96	227810	Fanconi-Bickel syndrome
SLC2A9	91	81	612076	Hypouricemia renal 2
SLC34A1	86	89	613388	Fanconi renotubular syndrome 2

SLC34A3	63	92	241530	Hypophosphatemic rickets with hypercalciuria
SLC3A1	133	92	220100	Cystinuria
SLC4A1	84	93	200	-
SLC4A4	74	41	604278	Renal tubular acidosis proximal with ocular abnormalities
SLC5A2	69	90	233100	Renal glucosuria
SLC6A19	96	87	234500	Hartnup disorder
SLC6A20	94	84	138500	Hyperglycinuria
SLC7A7	106	89	222700	Lysinuric protein intolerance
SLC7A9	83	92	220100	Cystinuria
SLC9A3R1	100	85	612287	Nephrolithiasis/osteoporosis hypophosphatemic 2
SMARCAL1	137	89	242900	Schimke immunoosseous dysplasia
SOX17	70	97	613674	Vesicoureteral reflux 3
STRA6	79	86	601186	Microphthalmia isolated with coloboma 8
STX16	104	91	603233	Pseudohypoparathyroidism type IB
TCTN1	112	89	200	-
TMEM216	78	88	608091	Joubert syndrome 2
TMEM237	99	91	614424	Joubert syndrome 14
TMEM67	104	95	216360	COACH syndrome
TRIM32	110	100	209900	Bardet-Biedl syndrome 11
TRPC6	97	72	603965	Glomerulosclerosis focal segmental 2
TTC8	107	91	209900	Bardet-Biedl syndrome 8
UMOD	88	85	609886	Glomerulocystic kidney disease with hyperuricemia and isosthenuria
UPK3A	57	88	191830	Renal adysplasia
VIPAS39	103	76	613404	Arthrogryposis renal dysfunction and cholestasis 2
VPS33B	121	89	208085	Arthrogryposis renal dysfunction and cholestasis 1
WDR19	122	94	614376	Asphyxiating thoracic dystrophy 5
WDR35	108	96	613610	Cranioectodermal dysplasia 2
WNK1	147	94	201300	Neuropathy hereditary sensory and autonomic type II
WNK4	111	93	614491	Pseudohypoaldosteronism type IIB
WNT4	130	95	158330	Mullerian aplasia and hyperandrogenism
WT1	63	91	194080	Denys-Drash syndrome
XPNPEP3	132	87	613159	Nephronophthisis-like nephropathy 1
ZNF423	124	98	614844	Joubert syndrome 19

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
Depth describes the average number of reads seen across 50 exomes
Coverage describes the average coverage of a gene across 50 exomes in percentiles
OMIM release used for OMIM disease identifiers and descriptions : 15 october 2013

Ad 1. OMIM identifier 200 signifies a gene without a current OMIM association

Ad 2. OMIM phenotype descriptions between {} signify risk factors