Oslo universitetssykehus
Avdeling for medisinsk genetikk

Nyresykdommer

Genpanel, versjon v01

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet, gjenkjennes (https://genome.ucsc.edu).

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ACE	2707	NM_000789.3		1-25	Renal tubular dysgenesis OMIM
ACTG2	145	NM_001615.3		2-9	Visceral myopathy OMIM
ACTN4	166	NM_004924.5	20-21	1-21	Glomerulosclerosis, focal segmental, 1 OMIM
ADAMTS13	1366	NM_139025.4		1-29	Thrombotic thrombocytopenic purpura, familial OMIM
ADCK4	19041	NM_024876.3		2-15	Nephrotic syndrome, type 9 OMIM
AGT	333	NM_000029.3		2-5	Renal tubular dysgenesis OMIM
AGTR1	336	NM_031850.3		3-4	Renal tubular dysgenesis OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
AGXT	341	NM_000030.2		1-11	Hyperoxaluria, primary, type 1 OMIM
AHI1	21575	NM_017651.4		3-28	Joubert syndrome 3 OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM
AMN	14604	NM_030943.3		1-12	Megaloblastic anemia-1, Norwegian type OMIM
ANKS6	26724	NM_173551.4		1-15	Nephronophthisis 16 OMIM
ANLN	14082	NM_018685.4		1-24	Focal segmental glomerulosclerosis 8 OMIM
APRT	626	NM_000485.2		1-5	Adenine phosphoribosyltransferase deficiency OMIM
AQP2	634	NM_000486.5		1-4	Diabetes insipidus, nephrogenic OMIM
ARHGDIA	678	NM_001185077.2		2-6	Nephrotic syndrome, type 8 OMIM
ARL6	13210	NM_177976.3		3-9	Bardet-Biedl syndrome 3 OMIM
ATP6V0A4	866	NM_020632.2		3-22	Renal tubular acidosis, distal, autosomal recessive OMIM
ATP6V1B1	853	NM_001692.3		1-14	Renal tubular acidosis with deafness OMIM
AVPR2	897	NM_000054.4		1-3	Diabetes insipidus, nephrogenic OMIM
B9D1	24123	NM_015681.4		1-7	?Meckel syndrome 9 OMIM
B9D2	28636	NM_030578.3		2-4	?Meckel syndrome 10 OMIM Joubert syndrome 34 OMIM
BBS1	966	NM_024649.4		1-17	Bardet-Biedl syndrome 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM
BBS12	26648	NM_152618.2		2	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3		1-17	Bardet-Biedl syndrome 2 OMIM
BBS4	969	NM_033028.4		1-16	Bardet-Biedl syndrome 4 OMIM
BBS5	970	NM_152384.2		1-12	Bardet-Biedl syndrome 5 OMIM
BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
BICC1	19351	NM_001080512.2		1-21	{Renal dysplasia, cystic, susceptibility to} OMIM
BMP4	1071	NM_001202.5		3-4	Microphthalmia, syndromic 6 OMIM
BSND	16512	NM_057176.2		1-4	Bartter syndrome, type 4a OMIM Sensorineural deafness with mild renal dysfunction OMIM
C1QA	1241	NM_015991.3		2-3	C1q deficiency OMIM
C1QB	1242	NM_000491.4		2-3	C1q deficiency OMIM
C1QC	1245	NM_172369.4		2-3	C1q deficiency OMIM
C3	1318	NM_000064.3		1-41	C3 deficiency OMIM {Hemolytic uremic syndrome, atypical, susceptibility to, 5} OMIM
C5orf42	25801	NM_023073.3		2-52	Joubert syndrome 17 OMIM Orofaciodigital syndrome VI OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CA2	1373	NM_000067.2		1-7	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis OMIM
CACNA1D	1391	NM_000720.3		1-49	Primary aldosteronism, seizures, and neurologic abnormalities OMIM
CACNA1H	1395	NM_021098.2		2-35	Hyperaldosteronism, familial, type IV OMIM
CASR	1514	NM_000388.3		2-7	Hypercalciuric hypercalcemia Hyperparathyroidism, neonatal OMIM Hypocalcemia, autosomal dominant OMIM Hypocalcemia, autosomal dominant, with Bartter syndrome OMIM Hypocalciuric hypercalcemia, type I OMIM
CC2D2A	29253	NM_001080522.2		3-38	COACH syndrome OMIM Meckel syndrome 6 OMIM
CCDC28B	28163	NM_024296.4		2-6	{Bardet-Biedl syndrome 1, modifier of} OMIM
CCDC41	17966	NM_016122.2		3-17	Nephronophthisis 18 OMIM
CD151	1630	NM_004357.4		3-9	Nephropathy with pretibial epidermolysis bullosa and deafness OMIM
CD2AP	14258	NM_012120.2		1-18	Glomerulosclerosis, focal segmental, 3 OMIM
CD46	6953	NM_002389.4	2-5	1-13	{Hemolytic uremic syndrome, atypical, susceptibility to, 2} OMIM
CEP164	29182	NM_014956.4		3-33	Nephronophthisis 15 OMIM
CEP290	29021	NM_025114.3	54	2-54	?Bardet-Biedl syndrome 14 OMIM Joubert syndrome 5 OMIM Meckel syndrome 4 OMIM Senior-Loken syndrome 6 OMIM
CFB	1037	NM_001710.5		1-18	{Hemolytic uremic syndrome, atypical, susceptibility to, 4} OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CFH	4883	NM_000186.3	8-10, 20-22	1-22	Complement factor H deficiency OMIM {Hemolytic uremic syndrome, atypical, susceptibility to, 1} OMIM
CFHR5	24668	NM_030787.3		1-10	Nephropathy due to CFHR5 deficiency OMIM
CFI	5394	NM_000204.4		1-13	{Hemolytic uremic syndrome, atypical, susceptibility to, 3} OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
CHRM3	1952	NM_000740.3		5	?Prune belly syndrome OMIM
CLCN5	2023	NM_000084.4		2-12	Dent disease OMIM Hypophosphatemic rickets OMIM Nephrolithiasis, type I OMIM Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis OMIM
CLDN16	2037	NM_006580.3		1-5	Hypomagnesemia 3, renal OMIM
CLDN19	2040	NM_148960.2		1-5	Hypomagnesemia 5, renal, with ocular involvement OMIM
CNNM2	103	NM_017649.4		1-8	Hypomagnesemia 6, renal OMIM
COL4A1	2202	NM_001845.5		1-52	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps OMIM
COL4A3	2204	NM_000091.4		1-52	Alport syndrome, autosomal dominant OMIM Alport syndrome, autosomal recessive OMIM Hematuria, benign familial OMIM
COL4A4	2206	NM_000092.4		2-48	Alport syndrome, autosomal recessive OMIM Hematuria, familial benign
COL4A5	2207	NM_000495.4		1-51	Alport syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
COQ2	25223	NM_015697.7		1-7	Coenzyme Q10 deficiency, primary, 1 OMIM {Multiple system atrophy, susceptibility to} OMIM
COQ6	20233	NM_182476.2		1-12	Coenzyme Q10 deficiency, primary, 6 OMIM
CRB2	18688	NM_173689.6		1-13	Focal segmental glomerulosclerosis 9 OMIM Ventriculomegaly with cystic kidney disease OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
CTNS	2518	NM_004937.2		3-12	Cystinosis, atypical nephropathic OMIM Cystinosis, late-onset juvenile or adolescent nephropathic OMIM Cystinosis, nephropathic OMIM
CUBN	2548	NM_001081.3	41-50, 61-67	1-67	Megaloblastic anemia-1, Finnish type OMIM
CYP24A1	2602	NM_000782.4		1-11	Hypercalcemia, infantile, 1 OMIM
DACT1	17748	NM_016651.5		1-4	?Townes-Brocks syndrome 2 OMIM
DCDC2	18141	NM_016356.4		1-10	Nephronophthisis 19 OMIM
DGKE	2852	NM_003647.2		2-12	Nephrotic syndrome, type 7 OMIM {Hemolytic uremic syndrome, atypical, susceptibility to, 7} OMIM
DMP1	2932	NM_004407.3		2-6	Hypophosphatemic rickets, AR OMIM
DSTYK	29043	NM_015375.2		1-13	Congenital anomalies of kidney and urinary tract 1 OMIM
DYNC2H1	2962	NM_001080463.1		1-90	Short-rib thoracic dysplasia 3 with or without polydactyly OMIM

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DZIP1L	26551	NM_173543.2		2-16	Polycystic kidney disease 5 OMIM
EHHADH	3247	NM_001966.3		1-7	?Fanconi renotubular syndrome 3 OMIM
EMP2	3334	NM_001424.5		2-5	Nephrotic syndrome, type 10 OMIM
ENPP1	3356	NM_006208.2		1-25	Arterial calcification, generalized, of infancy, 1 OMIM Hypophosphatemic rickets, autosomal recessive, 2 OMIM
EYA1	3519	NM_000503.5		3-18	Branchiootorenal syndrome 1, with or without cataracts OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FAM20A	23015	NM_017565.3		1-11	Amelogenesis imperfecta, type IG (enamel-renal syndrome) OMIM
FAN1	29170	NM_014967.4		2-14	Interstitial nephritis, karyomegalic OMIM
FGF20	3677	NM_019851.2		1-3	?Renal hypodysplasia/aplasia 2 OMIM
FGF23	3680	NM_020638.2		1-3	Hypophosphatemic rickets, autosomal dominant OMIM
FN1	3778	NM_212482.2		1-46	Glomerulopathy with fibronectin deposits 2 OMIM
FRAS1	19185	NM_025074.6		1-74	Fraser syndrome 1 OMIM
FREM1	23399	NM_144966.5		3-38	Bifid nose with or without anorectal and renal anomalies OMIM
FREM2	25396	NM_207361.5		1-24	Fraser syndrome 2 OMIM
FXYD2	4026	NM_001680.4		1-5	Hypomagnesemia 2, renal OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GANAB	4138	NM_198335.3		1-25	Polycystic kidney disease 3 OMIM
GATA3	4172	NM_001002295.1		2-6	Hypoparathyroidism, sensorineural deafness, and renal dysplasia OMIM
GLA	4296	NM_000169.2		1-7	Fabry disease OMIM
GLI3	4319	NM_000168.5		2-15	Pallister-Hall syndrome OMIM
GLIS2	29450	NM_032575.2		1-6	Nephronophthisis 7 OMIM
GREB1L	31042	NM_001142966.2		3-33	Renal hypodysplasia/aplasia 3 OMIM
GRHPR	4570	NM_012203.1		1-9	Hyperoxaluria, primary, type II OMIM
GRIP1	18708	NM_021150.3		1-24	Fraser syndrome 3 OMIM
HAAO	4796	NM_012205.2		1-10	Vertebral, cardiac, renal, and limb defects syndrome 1 OMIM
HNF1B	11630	NM_000458.3		1-9	Renal cysts and diabetes syndrome OMIM
HNF4A	5024	NM_175914.4		1-10	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young OMIM
HOGA1	25155	NM_138413.3		1-7	Hyperoxaluria, primary, type III OMIM
HPRT1	5157	NM_000194.2		1-9	HPRT-related gout OMIM Lesch-Nyhan syndrome OMIM
HPSE2	18374	NM_021828.4		1-12	Urofacial syndrome 1 OMIM
HSD11B2	5209	NM_000196.3		1-5	Apparent mineralocorticoid excess OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM

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IFT140	29077	NM_014714.3		3-31	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT172	30391	NM_015662.2		1-48	Short-rib thoracic dysplasia 10 with or without polydactyly OMIM
IFT43	29669	NM_052873.2		1-8	?Cranioectodermal dysplasia 3 OMIM
IFT80	29262	NM_020800.2		2-20	Short-rib thoracic dysplasia 2 with or without polydactyly OMIM
INF2	23791	NM_022489.3		2-22	Glomerulosclerosis, focal segmental, 5 OMIM
INPP5E	21474	NM_019892.5		1-10	Joubert syndrome 1 OMIM
INVS	17870	NM_014425.4		2-17	Nephronophthisis 2, infantile OMIM
IQCB1	28949	NM_001023570.3		3-15	Senior-Loken syndrome 5 OMIM
ITGA3	6139	NM_002204.3		1-25	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital OMIM
ITGA8	6144	NM_003638.2		1-30	Renal hypodysplasia/aplasia 1 OMIM
KAL1	6211	NM_000216.3	10-14	1-14	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1) OMIM
KANK2	29300	NM_001136191.2		3-13	Nephrotic syndrome 16 OMIM
KCNJ1	6255	NM_000220.4		1-2	Bartter syndrome, type 2 OMIM
KIF14	19181	NM_014875.2		2-30	?Meckel syndrome 12 OMIM
KIF7	30497	NM_198525.2		2-19	Joubert syndrome 12 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
KLHL3	6354	NM_017415.2		1-15	Pseudohypoaldosteronism, type IID OMIM
KYNU	6469	NM_003937.2		2-14	?Hydroxykynureninuria OMIM Vertebral, cardiac, renal, and limb defects syndrome 2 OMIM
LAMB2	6487	NM_002292.3		1-32	Nephrotic syndrome, type 5, with or without ocular abnormalities OMIM Pierson syndrome OMIM
LCAT	6522	NM_000229.1		1-6	Norum disease OMIM
LMX1B	6654	NM_002316.3		1-8	Nail-patella syndrome OMIM
LRIG2	20889	NM_014813.2		1-18	Urofacial syndrome 2 OMIM
LZTFL1	6741	NM_020347.3		1-10	Bardet-Biedl syndrome 17 OMIM
MAGED2	16353	NM_177433.2		2-12	Bartter syndrome, type 5, antenatal, transient OMIM
MAGI2	18957	NM_012301.3		1-22	Nephrotic syndrome 15 OMIM
MAPKBP1	29536	NM_014994.2		2-31	Nephronophthisis 20 OMIM
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AD OMIM Familial Mediterranean fever, AR OMIM
MKKS	7108	NM_018848.3		3-6	Bardet-Biedl syndrome 6 OMIM McKusick-Kaufman syndrome OMIM
MKS1	7121	NM_017777.3		1-18	Bardet-Biedl syndrome 13 OMIM Joubert syndrome 28 OMIM Meckel syndrome 1 OMIM
MYH9	7579	NM_002473.5		2-41	Epstein syndrome OMIM Fechtner syndrome OMIM
MYO1E	7599	NM_004998.3		1-28	Glomerulosclerosis, focal segmental, 6 OMIM

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NEK1	7744	NM_012224.2		2-34	Short-rib thoracic dysplasia 6 with or without polydactyly OMIM
NEK8	13387	NM_178170.2		1-15	?Nephronophthisis 9 OMIM Renal-hepatic-pancreatic dysplasia 2 OMIM
NOTCH2	7882	NM_024408.3	1-4	1-34	Alagille syndrome 2 OMIM Hajdu-Cheney syndrome OMIM
NPHP1	7905	NM_000272.3		1-20	Joubert syndrome 4 OMIM Nephronophthisis 1, juvenile OMIM Senior-Loken syndrome-1 OMIM
NPHP3	7907	NM_153240.4		1-27	Meckel syndrome 7 OMIM Nephronophthisis 3 OMIM Renal-hepatic-pancreatic dysplasia 1 OMIM
NPHP4	19104	NM_015102.4		2-30	Nephronophthisis 4 OMIM Senior-Loken syndrome 4 OMIM
NPHS1	7908	NM_004646.3		1-29	Nephrotic syndrome, type 1 OMIM
NPHS2	13394	NM_014625.3		1-8	Nephrotic syndrome, type 2 OMIM
NR3C1	7978	NM_001018077.1		2-9	Glucocorticoid resistance OMIM
NR3C2	7979	NM_000901.4		2-9	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy OMIM Pseudohypoaldosteronism type I, autosomal dominant OMIM
NUP107	29914	NM_020401.3		1-28	Nephrotic syndrome, type 11 OMIM
NUP205	18658	NM_015135.2		1-43	?Nephrotic syndrome, type 13 OMIM
NUP93	28958	NM_014669.4		2-22	Nephrotic syndrome, type 12 OMIM

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OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM Lowe syndrome OMIM Dent disease 2 OMIM Lowe syndrome OMIM
OFD1	2567	NM_003611.2		1-23	Joubert syndrome 10 OMIM Orofaciodigital syndrome I OMIM
PAX2	8616	NM_003987.4		1-11	Glomerulosclerosis, focal segmental, 7 OMIM Papillorenal syndrome OMIM
PBX1	8632	NM_002585.3		1-9	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay OMIM
PDE6D	8788	NM_002601.3		1-5	?Joubert syndrome 22 OMIM
PHEX	8918	NM_000444.5		1-22	Hypophosphatemic rickets, X-linked dominant OMIM
PKD1	9008	NM_001009944.2	1-33	1-46	Polycystic kidney disease 1 OMIM
PKD2	9009	NM_000297.3		1-15	Polycystic kidney disease 2 OMIM
PKHD1	9016	NM_138694.3		2-67	Polycystic kidney disease 4, with or without hepatic disease OMIM
PLCE1	17175	NM_016341.3		2-32	Nephrotic syndrome, type 3 OMIM
PMM2	9115	NM_000303.2		1-8	Congenital disorder of glycosylation, type Ia OMIM
PTPRO	9678	NM_030667.2		1-26	Nephrotic syndrome, type 6 OMIM
REN	9958	NM_000537.3		1-10	Hyperuricemic nephropathy, familial juvenile 2 OMIM Renal tubular dysgenesis OMIM
ROBO2	10250	NM_002942.4		1-26	Vesicoureteral reflux 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ROR2	10257	NM_004560.3		1-9	Robinow syndrome, autosomal recessive OMIM
RPGRIP1L	29168	NM_015272.4		2-27	COACH syndrome OMIM Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM
RRM2B	17296	NM_015713.4		1-9	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) OMIM
SCARB2	1665	NM_005506.3		1-12	Epilepsy, progressive myoclonic 4, with or without renal failure OMIM
SCNN1A	10599	NM_001038.5		2-13	Pseudohypoaldosteronism, type I OMIM
SCNN1B	10600	NM_000336.2		2-13	Liddle syndrome OMIM Pseudohypoaldosteronism, type I OMIM
SCNN1G	10602	NM_001039.3		2-13	Liddle syndrome OMIM Pseudohypoaldosteronism, type I OMIM
SDCCAG8	10671	NM_006642.4		1-18	Bardet-Biedl syndrome 16 OMIM Senior-Loken syndrome 7 OMIM
SGPL1	10817	NM_003901.3		2-15	Nephrotic syndrome 14 OMIM
SIX5	10891	NM_175875.4		1-3	Branchiootorenal syndrome 2 OMIM
SLC12A1	10910	NM_000338.2		2-27	Bartter syndrome, type 1 OMIM
SLC12A3	10912	NM_000339.2		1-26	Gitelman syndrome OMIM
SLC22A12	17989	NM_144585.3		1-10	Hypouricemia, renal OMIM
SLC2A9	13446	NM_020041.2		1-12	Hypouricemia, renal, 2 OMIM

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SLC34A1	11019	NM_003052.4		2-13	?Fanconi renotubular syndrome 2 OMIM Hypercalcemia, infantile, 2 OMIM Nephrolithiasis/osteoporosis, hypophosphatemic, 1 OMIM
SLC34A3	20305	NM_080877.2		2-13	Hypophosphatemic rickets with hypercalciuria OMIM
SLC36A2	18762	NM_181776.2		1-10	Hyperglycinuria OMIM
SLC3A1	11025	NM_000341.3		1-10	Cystinuria OMIM
SLC4A1	11027	NM_000342.3		2-20	Renal tubular acidosis, distal, AD OMIM Renal tubular acidosis, distal, AR OMIM
SLC4A4	11030	NM_003759.3		1-22	Renal tubular acidosis, proximal, with ocular abnormalities OMIM
SLC5A2	11037	NM_003041.3		1-14	Renal glucosuria OMIM
SLC6A19	27960	NM_001003841.2		1-12	Hyperglycinuria OMIM
SLC6A20	30927	NM_020208.3		1-11	Hyperglycinuria OMIM
SLC7A9	11067	NM_014270.4		2-13	Cystinuria OMIM
SLC9A3R1	11075	NM_004252.4		1-6	Nephrolithiasis/osteoporosis, hypophosphatemic, 2 OMIM
SMARCAL1	11102	NM_014140.3		3-18	Schimke immunoosseous dysplasia OMIM
SOX17	18122	NM_022454.3		1-2	Vesicoureteral reflux 3 OMIM
TBX18	11595	NM_001080508.2		1-8	Congenital anomalies of kidney and urinary tract 2 OMIM
TCTN2	25774	NM_024809.4		1-18	?Meckel syndrome 8 OMIM

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THBD	11784	NM_000361.2		1	{Hemolytic uremic syndrome, atypical, susceptibility to, 6} OMIM
TMEM107	28128	NM_183065.3		1-5	?Joubert syndrome 29 OMIM Meckel syndrome 13 OMIM
TMEM138	26944	NM_016464.4		2-5	Joubert syndrome 16 OMIM
TMEM216	25018	NM_001173990.2		1-5	Joubert syndrome 2 OMIM Meckel syndrome 2 OMIM
TMEM231	37234	NM_001077416.2		1-6	Joubert syndrome 20 OMIM Meckel syndrome 11 OMIM
TMEM237	14432	NM_001044385.2		1-12	Joubert syndrome 14 OMIM
TMEM67	28396	NM_153704.5		1-28	COACH syndrome OMIM Joubert syndrome 6 OMIM Meckel syndrome 3 OMIM Nephronophthisis 11 OMIM {Bardet-Biedl syndrome 14, modifier of} OMIM
TNXB	11976	NM_019105.6	17-18, 20-21, 23-24, 27-28, 32-44	2-44	Ehlers-Danlos syndrome, classic-like OMIM Vesicoureteral reflux 8 OMIM
TRAF3IP1	17861	NM_015650.3		1-17	Senior-Loken syndrome 9 OMIM
TRIM32	16380	NM_012210.3		2	?Bardet-Biedl syndrome 11 OMIM
TRPC6	12338	NM_004621.5		1-13	Glomerulosclerosis, focal segmental, 2 OMIM
TRPM6	17995	NM_017662.4		1-39	Hypomagnesemia 1, intestinal OMIM
TSC1	12362	NM_000368.4		3-23	Tuberous sclerosis-1 OMIM
TSC2	12363	NM_000548.4		2-42	Tuberous sclerosis-2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TTC21B	25660	NM_024753.4		1-29	Nephronophthisis 12 OMIM
TTC8	20087	NM_198309.3		2-15	Bardet-Biedl syndrome 8 OMIM
UMOD	12559	NM_003361.3		2-11	Glomerulocystic kidney disease with hyperuricemia and isosthenuria OMIM Hyperuricemic nephropathy, familial juvenile 1 OMIM Medullary cystic kidney disease 2 OMIM
VDR	12679	NM_001017535.1		4-11	Rickets, vitamin D-resistant, type IIA OMIM
VHL	12687	NM_000551.3		1-3	von Hippel-Lindau syndrome OMIM
VPS33B	12712	NM_018668.4		1-23	Arthrogryposis, renal dysfunction, and cholestasis 1 OMIM
WDR19	18340	NM_025132.3		1-36	?Cranioectodermal dysplasia 4 OMIM ?Short-rib thoracic dysplasia 5 with or without polydactyly OMIM Nephronophthisis 13 OMIM Senior-Loken syndrome 8 OMIM
WDR34	28296	NM_052844.3		1-9	Short-rib thoracic dysplasia 11 with or without polydactyly OMIM
WDR35	29250	NM_001006657.1		1-28	Cranioectodermal dysplasia 2 OMIM Short-rib thoracic dysplasia 7 with or without polydactyly OMIM
WDR60	21862	NM_018051.4		1-25	Short-rib thoracic dysplasia 8 with or without polydactyly OMIM
WDR73	25928	NM_032856.3		1-8	Galloway-Mowat syndrome 1 OMIM
WNK1	14540	NM_018979.3		1-28	Pseudohypoaldosteronism, type IIC OMIM
WNK4	14544	NM_032387.4		1-19	Pseudohypoaldosteronism, type IIB OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
WNT4	12783	NM_030761.4		1-5	?SERKAL syndrome OMIM
WT1	12796	NM_024426.4		1-10	Denys-Drash syndrome OMIM Frasier syndrome OMIM Meacham syndrome OMIM Mesothelioma, somatic OMIM Nephrotic syndrome, type 4 OMIM Wilms tumor, type 1 OMIM
XDH	12805	NM_000379.3		1-36	Xanthinuria, type I OMIM
XPNPEP3	28052	NM_022098.3		1-10	Nephronophthisis-like nephropathy 1 OMIM
ZMPSTE24	12877	NM_005857.4		1-10	Mandibuloacral dysplasia with type B lipodystrophy OMIM
ZNF423	16762	NM_015069.4		1-8	Joubert syndrome 19 OMIM Nephronophthisis 14 OMIM