Oslo universitetssykehus
Avdeling for medisinsk genetikk

Cystisk nyresykdom

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
AHI1	21575	NM_017651.4		3-28	Joubert syndrome 3 OMIM
ANKS6	26724	NM_173551.4		1-15	Nephronophthisis 16 OMIM
ARL6	13210	NM_177976.3		3-9	Bardet-Biedl syndrome 3 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
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
C5orf42	25801	NM_023073.3		2-52	Joubert syndrome 17 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
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
COL4A1	2202	NM_001845.5		1-52	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps OMIM
CRB2	18688	NM_173689.6		1-13	Ventriculomegaly with cystic kidney disease OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM

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DCDC2	18141	NM_016356.4		1-10	Nephronophthisis 19 OMIM
DYNC2H1	2962	NM_001080463.1		1-90	Short-rib thoracic dysplasia 3 with or without polydactyly OMIM
DZIP1L	26551	NM_173543.2		2-16	Polycystic kidney disease 5 OMIM
FAN1	29170	NM_014967.4		2-14	Interstitial nephritis, karyomegalic OMIM
GANAB	4138	NM_198335.3		1-25	Polycystic kidney disease 3 OMIM
GLIS2	29450	NM_032575.2		1-6	Nephronophthisis 7 OMIM
HNF1B	11630	NM_000458.3		1-9	Renal cysts and diabetes syndrome OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM
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
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
KIF14	19181	NM_014875.2		2-30	?Meckel syndrome 12 OMIM

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KIF7	30497	NM_198525.2		2-19	Joubert syndrome 12 OMIM
LZTFL1	6741	NM_020347.3		1-10	Bardet-Biedl syndrome 17 OMIM
MAPKBP1	29536	NM_014994.2		2-31	Nephronophthisis 20 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
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
OFD1	2567	NM_003611.2		1-23	Joubert syndrome 10 OMIM Orofaciodigital syndrome I OMIM
PDE6D	8788	NM_002601.3		1-5	?Joubert syndrome 22 OMIM
PKD1	9008	NM_001009944.2	1-33	1-46	Polycystic kidney disease 1 OMIM

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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
PMM2	9115	NM_000303.2		1-8	Congenital disorder of glycosylation, type Ia OMIM
RPGRIP1L	29168	NM_015272.4		2-27	COACH syndrome OMIM Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM
SDCCAG8	10671	NM_006642.4		1-18	Bardet-Biedl syndrome 16 OMIM Senior-Loken syndrome 7 OMIM
TCTN2	25774	NM_024809.4		1-18	?Meckel syndrome 8 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
TRAF3IP1	17861	NM_015650.3		1-17	Senior-Loken syndrome 9 OMIM
TRIM32	16380	NM_012210.3		2	?Bardet-Biedl syndrome 11 OMIM
TSC1	12362	NM_000368.4		3-23	Tuberous sclerosis-1 OMIM

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TSC2	12363	NM_000548.4		2-42	Tuberous sclerosis-2 OMIM
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
VHL	12687	NM_000551.3		1-3	von Hippel-Lindau syndrome 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
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
XPNPEP3	28052	NM_022098.3		1-10	Nephronophthisis-like nephropathy 1 OMIM
ZNF423	16762	NM_015069.4		1-8	Joubert syndrome 19 OMIM Nephronophthisis 14 OMIM