Oslo universitetssykehus
Avdeling for medisinsk genetikk

Renal tubulær sykdom

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
AGT	333	NM_000029.3		2-5	Renal tubular dysgenesis OMIM
AGTR1	336	NM_031850.3		3-4	Renal tubular dysgenesis OMIM
AQP2	634	NM_000486.5		1-4	Diabetes insipidus, nephrogenic 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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BSND	16512	NM_057176.2		1-4	Bartter syndrome, type 4a OMIM Sensorineural deafness with mild renal dysfunction OMIM
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
CCDC41	17966	NM_016122.2		3-17	Nephronophthisis 18 OMIM
CLCN5	2023	NM_000084.4		2-12	Dent disease OMIM Hypophosphatemic rickets 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
CTNS	2518	NM_004937.2		3-12	Cystinosis, atypical nephropathic OMIM Cystinosis, nephropathic OMIM
DMP1	2932	NM_004407.3		2-6	Hypophosphatemic rickets, AR OMIM
EHHADH	3247	NM_001966.3		1-7	?Fanconi renotubular syndrome 3 OMIM
ENPP1	3356	NM_006208.2		1-25	Arterial calcification, generalized, of infancy, 1 OMIM Hypophosphatemic rickets, autosomal recessive, 2 OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FGF23	3680	NM_020638.2		1-3	Hypophosphatemic rickets, autosomal dominant 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
GRHPR	4570	NM_012203.1		1-9	Hyperoxaluria, primary, type II OMIM
HOGA1	25155	NM_138413.3		1-7	Hyperoxaluria, primary, type III 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
KCNJ1	6255	NM_000220.4		1-2	Bartter syndrome, type 2 OMIM
KLHL3	6354	NM_017415.2		1-15	Pseudohypoaldosteronism, type IID OMIM
KYNU	6469	NM_003937.2		2-14	?Hydroxykynureninuria 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
OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM Lowe syndrome OMIM Dent disease 2 OMIM Lowe syndrome OMIM
PHEX	8918	NM_000444.5		1-22	Hypophosphatemic rickets, X-linked dominant 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
REN	9958	NM_000537.3		1-10	Hyperuricemic nephropathy, familial juvenile 2 OMIM Renal tubular dysgenesis 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
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
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
SLC34A1	11019	NM_003052.4		2-13	?Fanconi renotubular syndrome 2 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
SLC7A9	11067	NM_014270.4		2-13	Cystinuria OMIM
UMOD	12559	NM_003361.3		2-11	Hyperuricemic nephropathy, familial juvenile 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
WNK1	14540	NM_018979.3		1-28	Pseudohypoaldosteronism, type IIC OMIM
WNK4	14544	NM_032387.4		1-19	Pseudohypoaldosteronism, type IIB OMIM
XPNPEP3	28052	NM_022098.3		1-10	Nephronophthisis-like nephropathy 1 OMIM