Misdannelser i nyrer og urinveier

Genpanel, versjon v01

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

* 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 utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
ACE	2707	NM_000789.3		1-25	Renal tubular dysgenesis OMIM
ACTG2	145	NM_001615.3		2-9	Visceral myopathy OMIM
AGT	333	NM_000029.3		2-5	Renal tubular dysgenesis OMIM
AGTR1	336	NM_031850.3		3-4	Renal tubular dysgenesis OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome 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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
BSND	16512	NM_057176.2		1-4	Bartter syndrome, type 4a OMIM Sensorineural deafness with mild renal dysfunction OMIM
C5orf42	25801	NM_023073.3		2-52	Orofaciodigital syndrome VI OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
CHRM3	1952	NM_000740.3		5	?Prune belly syndrome OMIM
DACT1	17748	NM_016651.5		1-4	?Townes-Brocks syndrome 2 OMIM
DSTYK	29043	NM_015375.2		1-13	Congenital anomalies of kidney and urinary tract 1 OMIM
EYA1	3519	NM_000503.5		3-18	Branchiootorenal syndrome 1, with or without cataracts OMIM
FGF20	3677	NM_019851.2		1-3	?Renal hypodysplasia/aplasia 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
GATA3	4172	NM_001002295.1		2-6	Hypoparathyroidism, sensorineural deafness, and renal dysplasia OMIM
GLI3	4319	NM_000168.5		2-15	Pallister-Hall syndrome OMIM
GREB1L	31042	NM_001142966.2		3-33	Renal hypodysplasia/aplasia 3 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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
HNF1B	11630	NM_000458.3		1-9	Renal cysts and diabetes syndrome OMIM
HPSE2	18374	NM_021828.4		1-12	Urofacial syndrome 1 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
KYNU	6469	NM_003937.2		2-14	Vertebral, cardiac, renal, and limb defects syndrome 2 OMIM
LRIG2	20889	NM_014813.2		1-18	Urofacial syndrome 2 OMIM
NPHP3	7907	NM_153240.4		1-27	Meckel syndrome 7 OMIM Nephronophthisis 3 OMIM Renal-hepatic-pancreatic dysplasia 1 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
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
RPGRIP1L	29168	NM_015272.4		2-27	COACH syndrome OMIM Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
RRM2B	17296	NM_015713.4		1-9	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy) OMIM
SIX5	10891	NM_175875.4		1-3	Branchiootorenal syndrome 2 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
TNXB	11976	NM_019105.6	17-18, 20-21, 23-24, 27-28, 32-44	2-44	Vesicoureteral reflux 8 OMIM
VPS33B	12712	NM_018668.4		1-23	Arthrogryposis, renal dysfunction, and cholestasis 1 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	Short-rib thoracic dysplasia 7 with or without polydactyly OMIM
WNT4	12783	NM_030761.4		1-5	?SERKAL syndrome OMIM