Oslo universitetssykehus
Avdeling for medisinsk genetikk

Nyrestein og nefrokalsinose

Genpanel, versjon v02

* 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
ADCY10	21285	NM_001167749.2		3-30	{Hypercalciuria, absorptive, susceptibility to} OMIM
AGXT	341	NM_000030.2		1-11	Hyperoxaluria, primary, type 1 OMIM
APRT	626	NM_000485.3		1-5	Adenine phosphoribosyltransferase deficiency OMIM
ATP6V0A4	866	NM_020632.2		3-22	Distal renal tubular acidosis 3, with or without sensorineural hearing loss OMIM
ATP6V1B1	853	NM_001692.3		1-14	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CASR	1514	NM_001178065.1		2-7	Hyperparathyroidism, neonatal OMIM Hypocalcemia, autosomal dominant OMIM Hypocalcemia, autosomal dominant, with Bartter syndrome OMIM Hypocalciuric hypercalcemia, type I OMIM
CLCN5	2023	NM_001127899.3		3-15	Nephrolithiasis, type I OMIM
CLDN10	2033	NM_182848.3		1-5	HELIX syndrome OMIM
CLDN16	2037	NM_006580.3		1-5	Hypomagnesemia 3, renal OMIM
CLDN19	2040	NM_001123395.1		1-4	Hypomagnesemia 5, renal, with ocular involvement OMIM
CTNS	2518	NM_004937.2		3-12	Cystinosis, atypical nephropathic OMIM Cystinosis, nephropathic OMIM
CYP24A1	2602	NM_000782.4		1-11	Hypercalcemia, infantile, 1 OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FAM20A	23015	NM_001243746.2		3-12	Amelogenesis imperfecta, type IG (enamel-renal syndrome) OMIM
GRHPR	4570	NM_012203.2		1-9	Hyperoxaluria, primary, type II OMIM
HNF4A	5024	NM_175914.4		1-10	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young OMIM
HOGA1	25155	NM_001134670.1		1-3	Hyperoxaluria, primary, type III OMIM
HPRT1	5157	NM_000194.2		1-9	Hyperuricemia, HRPT-related OMIM Lesch-Nyhan syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
KCNJ1	6255	NM_153764.2		2	Bartter syndrome, type 2 OMIM
MAGED2	16353	NM_177433.3		2-12	Bartter syndrome, type 5, antenatal, transient OMIM
MOCOS	18234	NM_017947.4		1-15	Xanthinuria, type II OMIM
OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM
PRPS1	9462	NM_002764.4	7	1-7	Gout, PRPS-related OMIM
SLC12A1	10910	NM_001184832.2		2-27	Bartter syndrome, type 1 OMIM
SLC22A12	17989	NM_144585.3		1-10	Hypouricemia, renal OMIM
SLC26A1	10993	NM_134425.3		2-3	?Nephrolithiasis, calcium oxalate OMIM
SLC2A9	13446	NM_001001290.1		2-13	Hypouricemia, renal, 2 OMIM
SLC34A1	11019	NM_003052.5		2-13	Hypercalcemia, infantile, 2 OMIM Nephrolithiasis/osteoporosis, hypophosphatemic, 1 OMIM
SLC34A3	20305	NM_001177317.1		2-13	Hypophosphatemic rickets with hypercalciuria OMIM
SLC36A2	18762	NM_181776.2		1-10	Hyperglycinuria OMIM
SLC3A1	11025	NM_000341.4		1-10	Cystinuria OMIM
SLC4A1	11027	NM_000342.4		2-20	Distal renal tubular acidosis 1 OMIM Distal renal tubular acidosis 4 with hemolytic anemia OMIM
SLC6A19	27960	NM_001003841.2		1-12	Hyperglycinuria OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SLC6A20	30927	NM_020208.4		1-11	Hyperglycinuria OMIM
SLC7A9	11067	NM_014270.5		2-13	Cystinuria OMIM
SLC9A3R1	11075	NM_004252.5		1-6	Nephrolithiasis/osteoporosis, hypophosphatemic, 2 OMIM
XDH	12805	NM_000379.4		1-36	Xanthinuria, type I OMIM