Oslo universitetssykehus
Avdeling for medisinsk genetikk

Proteinuri og nefrotisk syndrome

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
ACTN4	166	NM_004924.5	20-21	1-21	Glomerulosclerosis, focal segmental, 1 OMIM
ADCK4	19041	NM_024876.3		2-15	Nephrotic syndrome, type 9 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
ANLN	14082	NM_018685.4		1-24	Focal segmental glomerulosclerosis 8 OMIM
ARHGDIA	678	NM_001185077.2		2-6	Nephrotic syndrome, type 8 OMIM
C3	1318	NM_000064.3		1-41	C3 deficiency OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CD2AP	14258	NM_012120.2		1-18	Glomerulosclerosis, focal segmental, 3 OMIM
CFH	4883	NM_000186.3	8-10, 20-22	1-22	Complement factor H deficiency OMIM
CLCN5	2023	NM_000084.4		2-12	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis OMIM
COQ2	25223	NM_015697.7		1-7	Coenzyme Q10 deficiency, primary, 1 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
CTNS	2518	NM_004937.2		3-12	Cystinosis, late-onset juvenile or adolescent nephropathic OMIM
CUBN	2548	NM_001081.3	41-50, 61-67	1-67	Megaloblastic anemia-1, Finnish type OMIM
DGKE	2852	NM_003647.2		2-12	Nephrotic syndrome, type 7 OMIM
EMP2	3334	NM_001424.5		2-5	Nephrotic syndrome, type 10 OMIM
GLA	4296	NM_000169.2		1-7	Fabry disease OMIM
INF2	23791	NM_022489.3		2-22	Glomerulosclerosis, focal segmental, 5 OMIM
ITGA3	6139	NM_002204.3		1-25	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital OMIM
KANK2	29300	NM_001136191.2		3-13	Nephrotic syndrome 16 OMIM
LAMB2	6487	NM_002292.3		1-32	Nephrotic syndrome, type 5, with or without ocular abnormalities OMIM Pierson syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
LCAT	6522	NM_000229.1		1-6	Norum disease OMIM
LMX1B	6654	NM_002316.3		1-8	Nail-patella syndrome OMIM
MAGI2	18957	NM_012301.3		1-22	Nephrotic syndrome 15 OMIM
MEFV	6998	NM_000243.2		1-10	Familial Mediterranean fever, AD OMIM Familial Mediterranean fever, AR OMIM
MYH9	7579	NM_002473.5		2-41	Fechtner syndrome OMIM
MYO1E	7599	NM_004998.3		1-28	Glomerulosclerosis, focal segmental, 6 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
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
OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM Dent disease 2 OMIM
PAX2	8616	NM_003987.4		1-11	Glomerulosclerosis, focal segmental, 7 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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SCARB2	1665	NM_005506.3		1-12	Epilepsy, progressive myoclonic 4, with or without renal failure OMIM
SGPL1	10817	NM_003901.3		2-15	Nephrotic syndrome 14 OMIM
SMARCAL1	11102	NM_014140.3		3-18	Schimke immunoosseous dysplasia OMIM
TRPC6	12338	NM_004621.5		1-13	Glomerulosclerosis, focal segmental, 2 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
WDR73	25928	NM_032856.3		1-8	Galloway-Mowat syndrome 1 OMIM
WT1	12796	NM_024426.4		1-10	Nephrotic syndrome, type 4 OMIM
ZMPSTE24	12877	NM_005857.4		1-10	Mandibuloacral dysplasia with type B lipodystrophy OMIM