Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for torakalt aortaaneurisme og aortadisseksjon

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
ABL1	76	NM_005157.6		1-11	Congenital heart defects and skeletal malformations syndrome OMIM
ACTA2	130	NM_001141945.1		2-9	Aortic aneurysm, familial thoracic 6 OMIM Moyamoya disease 5 OMIM Multisystemic smooth muscle dysfunction syndrome OMIM
BGN	1044	NM_001711.6		2-8	Meester-Loeys syndrome OMIM
COL1A1	2197	NM_000088		1-51	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1 OMIM Ehlers-Danlos syndrome, arthrochalasia type, 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
COL1A2	2198	NM_000089		1-52	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2 OMIM Ehlers-Danlos syndrome, arthrochalasia type, 2 OMIM Ehlers-Danlos syndrome, cardiac valvular type OMIM
COL3A1	2201	NM_000090.3		1-51	Ehlers-Danlos syndrome, vascular type OMIM Polymicrogyria with or without vascular-type EDS OMIM
COL5A1	2209	NM_001278074		1-66	Ehlers-Danlos syndrome, classic type, 1 OMIM Fibromuscular dysplasia, multifocal OMIM
COL5A2	2210	NM_000393.3		1-54	Ehlers-Danlos syndrome, classic type, 2 OMIM
EFEMP2	3219	NM_016938.4		2-11	Cutis laxa, autosomal recessive, type IB OMIM
ELN	3327	NM_000501		1-33	Cutis laxa, autosomal dominant OMIM
FBLN5	3602	NM_006329		1-11	?Cutis laxa, autosomal dominant 2 OMIM Cutis laxa, autosomal recessive, type IA OMIM
FBN1	3603	NM_000138.4		2-66	Marfan lipodystrophy syndrome OMIM Marfan syndrome OMIM
FBN2	3604	NM_001999.3		1-65	Contractural arachnodactyly, congenital OMIM
FKBP14	18625	NM_017946		1-4	Ehlers-Danlos syndrome, kyphoscoliotic type, 2 OMIM
FLNA	3754	NM_001456		2-47	Heterotopia, periventricular, 1 OMIM
FOXE3	3808	NM_012186.2		1	{Aortic aneurysm, familial thoracic 11, susceptibility to} OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
IPO8	9853	NM_006390.3		1-25	VISS syndrome OMIM
LOX	6664	NM_002317		1-7	Aortic aneurysm, familial thoracic 10 OMIM
LTBP3	6716	NM_001130144.2		1-28	Dental anomalies and short stature OMIM
MFAP5	29673	NM_003480.4		2-10	Aortic aneurysm, familial thoracic 9 OMIM
MYH11	7569	NM_001040114		2-42	Aortic aneurysm, familial thoracic 4 OMIM
MYLK	7590	NM_053025	13-18	4-34	Aortic aneurysm, familial thoracic 7 OMIM
NOTCH1	7881	NM_017617.3		1-34	Aortic valve disease 1 OMIM
PLOD1	9081	NM_000302		1-19	Ehlers-Danlos syndrome, kyphoscoliotic type, 1 OMIM
PRKG1	9414	NM_001098512.2		1-18	Aortic aneurysm, familial thoracic 8 OMIM
SKI	10896	NM_003036		1-7	Shprintzen-Goldberg syndrome OMIM
SLC2A10	13444	NM_030777.3		1-5	Arterial tortuosity syndrome OMIM
SMAD2	6768	NM_001003652		2-11	Loeys-Dietz syndrome 5
SMAD3	6769	NM_005902		1-9	Loeys-Dietz syndrome 3 OMIM
SMAD4	6770	NM_005359.6		2-12	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome OMIM
SMAD6	6772	NM_005585.4		1-4	Aortic valve disease 2 OMIM
TGFB2	11768	NM_003238.6		1-7	Loeys-Dietz syndrome 4 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TGFB3	11769	NM_003239.3		1-7	Loeys-Dietz syndrome 5 OMIM
TGFBR1	11772	NM_004612.3		1-9	Loeys-Dietz syndrome 1 OMIM
TGFBR2	11773	NM_003242		1-7	Loeys-Dietz syndrome 2 OMIM
THSD4	25835	NM_024817.2		1-17	Aortic aneurysm, familial thoracic PubMed