SNiPAcard

Block annotations

Block info

genomic range	chr9:136,194,812-136,284,450
block size	89,639 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.780 [-3.93 – 0.701]	**gene(s) hit or close-by**	ADAMTS13, AL593848.2, MED22, REXO4, RP11-244N20.7, RPL7A, SNORD24, SNORD36A, SNORD36B, SNORD36C, SURF1, SURF2, SURF4, SURF6, Y_RNA
phastCons	μ = 0.021 [0 – 0.275]	**eQTL gene(s)**	ADAMTS13, CACFD1, MED22, REXO4, RP11-244N20.7, SLC2A6, SURF1, SURF2, SURF6
GERP++	μ = -1.493 [-6.07 – 2.05]	**potentially regulated gene(s)**	AK8, EEF1A1P5, GTF3C5, REXO4, REXO4, SARDH, SURF1, SURF1, SURF2, SURF2
CADD score	μ = 3.308 [0.026 – 12.47]	**disease gene(s)**	ADAMTS13, SURF1, SARDH,

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Stroke	2.45×10-4	dbGaP	pha002886	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	benign	ClinVar	RCV000128344.1	1

Disease gene annotation

gene	trait	source DB	source entry/link
ADAMTS13	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL	OMIM	MIM:274150
SURF1	LEIGH SYNDROME	OMIM	MIM:256000
SARDH	SARCOSINEMIA	OMIM	MIM:268900
SURF1	Complex IV deficiency	DECIPHER	MIM:220110
SURF1	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
	Complex IV deficiency	DECIPHER	MIM:220110
	Leigh Syndrome (nuclear DNA mutation)	DECIPHER	MIM:256000
ADAMTS13	Congenital thrombotic thrombocytopenic purpura due to ADAMTS-13 deficiency	OrphaNet	OrphaNet:93583
SURF1	Fatal infantile cytochrome C oxidase deficiency	OrphaNet	OrphaNet:1561
SURF1	Leigh syndrome with cardiomyopathy	OrphaNet	OrphaNet:70474
SURF1	Leigh syndrome with leukodystrophy	OrphaNet	OrphaNet:255241
SURF1	SURF1-related Charcot-Marie-Tooth disease type 4	OrphaNet	OrphaNet:391351
SARDH	SARCOSINEMIA	OrphaNet	OrphaNet:3129

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SURF6	ENST00000372022	ILMN_1778032	skin	5.13×10-20 (p-value)	MuTHER consortium	10
SURF6	ENST00000468290		blood	1.68×10-33 (p-value)	MuTHER consortium	10

			adipocyte	1.06×10-20 (p-value)	MuTHER consortium	10
SURF1	?	ENSG00000148290	atrial appendage	8.22×10-10 (p-value)	GTEx Portal V6	23
REXO4	?	ENSG00000148300	transformed fibroblasts	4.52×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	transformed fibroblasts	1.04×10-14 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	blood	4.49×10-40 (p-value)	GTEx Portal V6	23
RP11-244N20.7	?	ENSG00000230064	blood	1.95×10-9 (p-value)	GTEx Portal V6	23
MED22	?	ENSG00000148297	blood	9.70×10-25 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	blood	4.29×10-6 (p-value)	GTEx Portal V6	1
SURF6	?	ENSG00000148296	breast	5.95×10-8 (p-value)	GTEx Portal V6	16
SURF1	?	ENSG00000148290	breast	6.16×10-17 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	blood	1.94×10-4 (q-value)	SeeQTL DB (HapMap)	10
SURF1	?	ENSG00000148290	blood	1.07×10-12 (q-value)	SeeQTL DB (HapMap)	10
SURF1	?	ENSG00000148290	EBV lymphocytes	3.39×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	sun exposed skin	7.05×10-27 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	sun exposed skin	1.09×10-25 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	sun exposed skin	1.22×10-8 (p-value)	GTEx Portal V6	12
SURF6	?	ENSG00000148296	unexposed skin	1.86×10-19 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	unexposed skin	3.05×10-12 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	aorta	7.43×10-10 (p-value)	GTEx Portal V6	22
SURF6	?	ENSG00000148296	left ventricle	4.00×10-10 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	left ventricle	2.51×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	subcutaneous adipocytes	4.51×10-19 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	subcutaneous adipocytes	2.85×10-30 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	subcutaneous adipocytes	2.72×10-9 (p-value)	GTEx Portal V6	18
SURF2	?	ENSG00000148291	subcutaneous adipocytes	2.52×10-6 (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	caudate basal ganglia	6.03×10-10 (p-value)	GTEx Portal V6	21
ADAMTS13	?	ENSG00000160323	caudate basal ganglia	9.38×10-8 (p-value)	GTEx Portal V6	11
SURF6	?	ENSG00000148296	visceral adipocytes	1.99×10-12 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	visceral adipocytes	3.18×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	stomach	3.98×10-9 (p-value)	GTEx Portal V6	22
ADAMTS13	?	ENSG00000160323	cortex	6.77×10-8 (p-value)	GTEx Portal V6	17
SURF1	?	ENSG00000148290	cortex	2.90×10-9 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	tibial nerve	7.30×10-13 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	tibial nerve	2.93×10-13 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	tibial nerve	4.00×10-7 (p-value)	GTEx Portal V6	15
SURF6	?	ENSG00000148296	esophagus mucosa	4.21×10-14 (p-value)	GTEx Portal V6	23

SURF6	?	ENSG00000148296	esophagus mucosa	4.21×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	esophagus mucosa	1.14×10-18 (p-value)	GTEx Portal V6	23
CACFD1	?	ENSG00000160325	esophagus mucosa	3.18×10-7 (p-value)	GTEx Portal V6	16
SURF1	?	ENSG00000148290	anterior cingulate cortex	3.43×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	testis	3.25×10-14 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	frontal cortex	1.25×10-10 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	frontal cortex	2.35×10-8 (p-value)	GTEx Portal V6	2
SURF6	?	ENSG00000148296	pancreas	1.13×10-10 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	muscularis mucosae	2.30×10-16 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	muscularis mucosae	1.47×10-7 (p-value)	GTEx Portal V6	13
SURF6	?	ENSG00000148296	lung	2.37×10-15 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	lung	4.25×10-16 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	lung	4.53×10-6 (p-value)	GTEx Portal V6	1
SURF6	?	ENSG00000148296	tibial artery	2.73×10-11 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	tibial artery	4.09×10-9 (p-value)	GTEx Portal V6	18
SURF6	?	ENSG00000148296	thyroid	9.98×10-10 (p-value)	GTEx Portal V6	22
CACFD1	?	ENSG00000160325	thyroid	2.38×10-11 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	thyroid	4.92×10-14 (p-value)	GTEx Portal V6	23
ADAMTS13	?	ENSG00000160323	thyroid	2.24×10-7 (p-value)	GTEx Portal V6	13
SURF6	?	ENSG00000148296	transverse colon	1.30×10-10 (p-value)	GTEx Portal V6	23
SURF1	?	ENSG00000148290	transverse colon	6.90×10-14 (p-value)	GTEx Portal V6	23
SURF6	ENST00000372022	ILMN_1778032	monocyte	1.38×10-191 (p-value)	Zeller et al.	3
SURF6	ENST00000468290					
MED22	ENST00000614493	ILMN_1792860	monocyte	2.23×10-20 (p-value)	Zeller et al.	2
MED22	ENST00000610888					
MED22	ENST00000614493	ILMN_1697218	monocyte	2.87×10-60 (p-value)	Zeller et al.	2
MED22	ENST00000610672					
MED22	ENST00000343730					
MED22	ENST00000610888					
?	?	ILMN_1663407	monocyte	2.76×10-21 (p-value)	Zeller et al.	1
SURF6	?	ENSG00000148296	terminal ileum	1.93×10-7 (p-value)	GTEx Portal V6	8
SURF6	?	ENSG00000148296	spleen	2.60×10-15 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	sigmoid colon	1.89×10-8 (p-value)	GTEx Portal V6	14
SURF6	?	ENSG00000148296	gastroesophageal junction	5.21×10-11 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	adrenal gland	2.40×10-8 (p-value)	GTEx Portal V6	18
SURF1	?	ENSG00000148290	cerebellar hemisphere	1.08×10-9 (p-value)	GTEx Portal V6	23
SURF6	?	ENSG00000148296	cerebellum	8.00×10-8 (p-value)	GTEx Portal V6	21
SURF1	?	ENSG00000148290	cerebellum	3.12×10-15 (p-value)	GTEx Portal V6	23

SURF1	?	ENSG00000148290	hypothalamus	1.36×10^{-8} (p-value)	GTEx Portal V6	14
SURF1	?	ENSG00000148290	prostate	1.35×10^{-7} (p-value)	GTEx Portal V6	6
MED22	ENST00000614493	ILMN_1697218	monocyte	3.91×10^{-19} (p-value)	Fairfax et al.	2
MED22	ENST00000610672		b-cell	2.08×10^{-16} (p-value)	Fairfax et al.	2
MED22	ENST00000343730					
MED22	ENST00000610888					
SURF6	ENST00000372022	ILMN_1778032	monocyte	1.29×10^{-43} (p-value)	Fairfax et al.	2
SURF6	ENST00000468290		b-cell	1.11×10^{-17} (p-value)	Fairfax et al.	2
?	?	ILMN_1663407	monocyte	6.11×10^{-5} (p-value)	Fairfax et al.	2
			b-cell	1.67×10^{-5} (p-value)		
SLC2A6	ENST00000371899	ILMN_1778321	blood	6.13×10^{-7} (p-value)	Westra et al.	5
SLC2A6	ENST00000485978					
SLC2A6	ENST00000371897					
SURF1	?	ENSG00000148290	skeletal muscle	4.20×10^{-7} (p-value)	GTEx Portal V6	3
SURF1	?	ENSG00000148290	hippocampus	1.61×10^{-7} (p-value)	GTEx Portal V6	5
SURF1	?	ENSG00000148290	brain	1.67×10^{-2} (q-value)	SeeQTL DB (Myers et al.)	1
ADAMTS13	?	ENSG00000160323	pituitary	1.95×10^{-6} (p-value)	GTEx Portal V6	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
MED22	ENST00000614493	ILMN_1792860	chr9	monocyte	5.32×10^{-23} (p-value)	Zeller et al.	1
MED22	ENST00000610888						
MED22	ENST00000614493	ILMN_1697218	chr9	monocyte	7.47×10^{-66} (p-value)	Zeller et al.	1
MED22	ENST00000610672						
MED22	ENST00000343730						
MED22	ENST00000610888						
?	?	ILMN_1663407	chr9	monocyte	9.27×10^{-22} (p-value)	Zeller et al.	2

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000060505		SURF1
ENCP00000060506		SURF2
		SURF1
		SURF1
ENCP00000060514		REXO4

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000558368	1	ENCP00000060532	SARDH
		ENCP00000060469	AK8
ENCE00000558463	1	ENCP00000060478	EEF1A1P5
		ENCP00000060479	GTF3C5

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors

element id	variant(s)	tissue/cell	factors
ENSR00000049921 (promoter)	1	embryonic stem cell (H1ESC)	H3K27ac, H3K36me3, CTCF, PolII, Rad21, Egr1, Gabp, Sin3Ak20, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K4me2, H3K4me3, DNase1
		blood (K562)	DNase1, H3K4me2, PolII, ELF1, TAF1, HEY1, Cmyc, Gabp, H3K9ac, Max, Egr1, Yy1, H3K4me3
		skin (NHDF-AD)	H3K9ac, H3K4me2, DNase1, H3K4me3
		muscle (HSMM)	H3K9ac, H3K4me2, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, Gabp, Yy1, PolII, TAF1, ELF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, Egr1, Yy1, Gabp, ELF1, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	CTCF, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K4me3, H3K36me3
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K4me1
		breast (HMEC)	DNase1, CTCF, H3K4me2, H3K4me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, H3K4me2, DNase1
		endothelium (HUVEC)	H3K36me3, H3K4me3, PolII, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K36me3
ENSR00000049926 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K79me2, H3K36me3
		blood (K562)	H3K36me3, H4K20me1, H3K79me2, PolII
		muscle (HSMM)	H3K36me3, H4K20me1, H3K79me2
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		blood (GM12878)	H3K36me3, H3K4me2, H3K27ac, PolII, H3K79me2, H3K4me3
		nervous (NH-A)	H4K20me1, H3K36me3
		skin (NHEK)	H3K4me1, H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3, H3K4me2, H4K20me1
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001475646 (promoter)	2	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Egr1, Gabp, Jund, TAF7, H3K9ac, H3K27ac, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K4me2, H2AZ, DNase1
		blood (K562)	H3K27ac, Max, E2F6, H3K9ac, H3K4me3, DNase1, H3K4me2, PolII, TAF1, H2AZ, HEY1, H3K79me2
		skin (NHDF-AD)	H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K79me2, H2AZ, H3K4me3, H3K27ac, DNase1, H3K9ac, H3K4me2
		liver (HepG2)	DNase1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, H3K4me2, H3K4me1, Gabp, PolII, H3K79me2, TAF1, ELF1
		lung (IMR90)	DNase1, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		skin (NHEK)	CTCF, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1, H3K36me3
		breast (HMEC)	H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3, H3K36me3, H4K20me1, H3K9ac
		endothelium (HUVEC)	H3K36me3, PolII, DNase1, H3K27ac, H3K9ac, H3K4me3, Max, H3K4me2
		A549	H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3, H3K4me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ADAMTS13	upstream gene variant	2670	ENST00000371916	?	ENSP00000360984	1
ADAMTS13	upstream gene variant	2995	ENST00000474918	?	ENSP00000435305	1

ADAMTS13	upstream gene variant	2994	ENST00000495234	?	ENSP00000435274 1
ADAMTS13	upstream gene variant	2994	ENST00000371911	?	ENSP00000360979 1
ADAMTS13	upstream gene variant	2994	ENST00000355699	NM_139027.4	ENSP00000347927 1
ADAMTS13	upstream gene variant	2670	ENST00000371929	NM_139025.4	ENSP00000360997 1
ADAMTS13	upstream gene variant	2994	ENST00000356589	NM_139026.4	ENSP00000348997 1
AL593848.2	downstream gene variant, upstream gene variant	527	ENST00000626556	?	? 5
MED22	downstream gene variant, upstream gene variant	4032	ENST00000610672	?	ENSP00000482438 3
MED22	downstream gene variant, upstream gene variant	3660	ENST00000471524	?	? 3
MED22	downstream gene variant, upstream gene variant	1967	ENST00000343730	NM_133640.4	ENSP00000342343 3
MED22	upstream gene variant	3612	ENST00000494177	?	ENSP00000420815 2
MED22	downstream gene variant, upstream gene variant	893	ENST00000482295	?	? 3
MED22	upstream gene variant	3604	ENST00000457204	?	ENSP00000404178 2
MED22	downstream gene variant, upstream gene variant	1967	ENST00000610888	NM_181491.2	ENSP00000478773 3
MED22	downstream gene variant, upstream gene variant	3618	ENST00000371999	?	ENSP00000361067 3
MED22	upstream gene variant	3713	ENST00000446777	?	ENSP00000394781 2
MED22	downstream gene variant, upstream gene variant	1968	ENST00000614493	?	ENSP00000481493 3
REXO4	downstream gene variant, upstream gene variant	187	ENST00000371935	NM_001279349.1	ENSP00000361003 3
REXO4	downstream gene variant, upstream gene variant	190	ENST00000371942	NM_001279350.1, NM_001279351.1, NM_020385.3	ENSP00000361010 3
REXO4	downstream gene variant, upstream gene variant	1134	ENST00000454825	?	ENSP00000394229 3
REXO4	downstream gene variant, upstream gene variant	1371	ENST00000478037	?	? 2
REXO4	downstream gene variant, upstream gene variant	1376	ENST00000494045	?	? 2
REXO4	downstream gene variant, upstream gene variant	733	ENST00000445916	?	ENSP00000391534 3
RP11-244N20.7	upstream gene variant	52	ENST00000439130	?	? 1
RPL7A	upstream gene variant, downstream gene variant	1040	ENST00000496554	?	? 5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000315731	?	ENSP00000361071 5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000323345	NM_000972.2	ENSP00000361076 5
RPL7A	upstream gene variant, downstream gene variant	363	ENST00000468019	?	? 5
RPL7A	upstream gene variant, downstream gene variant	309	ENST00000463740	?	? 5
RPL7A	upstream gene variant, downstream gene variant	1352	ENST00000485706	?	? 4
RPL7A	upstream gene variant, downstream gene variant	1417	ENST00000426651	?	ENSP00000416638 4
RPL7A	upstream gene variant, downstream gene variant	350	ENST00000489392	?	? 5
RPL7A	downstream gene variant	322	ENST00000492798	?	? 4
SNORD24	upstream gene variant, downstream gene variant	2265	ENST00000383884	?	? 4
SNORD36A	upstream gene variant, downstream gene variant	1207	ENST00000362874	?	? 4
SNORD36B	upstream gene variant, downstream gene variant	1567	ENST00000363961	?	? 4
SNORD36C	downstream gene variant	823	ENST00000516733	?	? 4

SURF1	downstream gene variant, upstream gene variant	170	ENST00000437995 ?	?	3
SURF1	downstream gene variant, upstream gene variant	5	ENST00000371974 NM_003172.3	ENSP00000361042	3
SURF1	downstream gene variant	470	ENST00000463965 ?	?	4
SURF1	downstream gene variant, upstream gene variant	105	ENST00000495952 ?	?	5
SURF1	downstream gene variant, upstream gene variant	5	ENST00000615505 NM_001280787.1	ENSP00000482067	3
SURF2	upstream gene variant	558	ENST00000486887 ?	?	3
SURF2	upstream gene variant	88	ENST00000495524 ?	?	5
SURF2	upstream gene variant	62	ENST00000371964 NM_017503.4, NM_001278928.1	ENSP00000361032	5
SURF4	downstream gene variant	4960	ENST00000618229 NM_001280790.1	ENSP00000483216	1
SURF4	downstream gene variant	4960	ENST00000545297 NM_001280792.1	ENSP00000446061	1
SURF4	downstream gene variant	4960	ENST00000613129 NM_001280791.1	ENSP00000481666	1
SURF4	downstream gene variant	4959	ENST00000371989 NM_033161.3, NM_001280788.1	ENSP00000361057	1
SURF6	downstream gene variant, upstream gene variant	808	ENST00000468290 ?	?	7
SURF6	downstream gene variant, upstream gene variant	799	ENST00000372022 NM_006753.5	ENSP00000361092	2
Y_RNA	downstream gene variant, upstream gene variant	1114	ENST00000364581 ?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ADAMTS13	ENST00000485925	?	?	2
MED22	ENST00000494177	?	ENSP00000420815	1
MED22	ENST00000610888	NM_181491.2	ENSP00000478773	1
MED22	ENST00000610672	?	ENSP00000482438	1
MED22	ENST00000446777	?	ENSP00000394781	1
MED22	ENST00000457204	?	ENSP00000404178	1
MED22	ENST00000471524	?	?	1
MED22	ENST00000614493	?	ENSP00000481493	1
MED22	ENST00000371999	?	ENSP00000361067	1
MED22	ENST00000343730	NM_133640.4	ENSP00000342343	1
REXO4	ENST00000494045	?	?	1
REXO4	ENST00000478037	?	?	1
REXO4	ENST00000371935	NM_001279349.1	ENSP00000361003	2
REXO4	ENST00000454825	?	ENSP00000394229	2
REXO4	ENST00000371942	NM_001279350.1, NM_001279351.1, NM_020385.3	ENSP00000361010	2
SURF1	ENST00000371974	NM_003172.3	ENSP00000361042	2
SURF1	ENST00000615505	NM_001280787.1	ENSP00000482067	2
SURF1	ENST00000437995	?	?	2
SURF4	ENST00000545297	NM_001280792.1	ENSP00000446061	4
SURF4	ENST00000485435	NM_001280789.1	ENSP00000419853	4

SURF4	ENST00000371989	NM_033161.3, NM_001280788.1	ENSP00000361057	4
SURF4	ENST00000618229	NM_001280790.1	ENSP00000483216	4
SURF4	ENST00000371991	?	ENSP00000361059	4
SURF4	ENST00000371982	?	?	4
SURF4	ENST00000613129	NM_001280791.1	ENSP00000481666	4
SURF4	ENST00000467910	?	?	4
SURF6	ENST00000372022	NM_006753.5	ENSP00000361092	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MED22	ENST00000610672	?	ENSP00000482438	1
SURF6	ENST00000372022	NM_006753.5	ENSP00000361092	5

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SURF1	ENST00000463965	?	1