Locus 130 (rs13063312)

SNiPAcard

Block annotations

Block info

genomic range	chr3:48,661,985-48,765,168
block size	103,184 bp
variant count	75 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.176 [-2.807 – 4.885]	gene(s) hit or close-by	AC141002.1, CELSR3, CELSR3-AS1, IP6K2, MIR4793, MIR6824, NCKIPSD, RP11-572O6.1, SLC26A6, TMEM89
phastCons	μ = 0.103 [0 – 1]	eQTL gene(s)	ATRIP, C3orf62, CCDC36, CCDC71, CELSR3, IP6K2, KLHDC8B, MIR5193, NCKIPSD, NICN1, PFKFB4, RBM6, RP11-3B7.1, SLC26A6, TMEM89, TREX1, TREX1, UBA7, USP4, WDR6
GERP++	μ = -0.679 [-8.07 – 5.67]	potentially regulated gene(s)	ARIH2, ATRIP, CCDC51, CDC25A, COL7A1, COL7A1, DALRD3, IP6K2, NCKIPSD, PLXNB1, PRKAR2A, QARS, QRICH1, SHISA5, SLC25A20, SLC26A6, SPINK8, WDR6, ZNF589
CADD score	μ = 5.844 [0.048 – 22.1]	disease gene(s)	KLHDC8B, TREX1, TREX1, QARS, COL7A1, SLC25A20, ATRIP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood pressure	2.57×10^{-5}	dbGaP	pha003048	2
Blood pressure	9.40×10^{-6}	dbGaP	pha003047	3
Blood pressure	2.30×10^{-5}	dbGaP	pha003046	2
Blood pressure	2.52×10^{-5}	dbGaP	pha003045	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CM086000	1

Disease gene annotation

gene	trait	source DB	source entry/link
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760
COL7A1	NAIL DISORDER, NONSYNDROMIC CONGENITAL, 8	OMIM	MIM:607523
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, PRETIBIAL	OMIM	MIM:131850
COL7A1	EPIDERMOLYSIS BULLOSA PRURIGINOSA	OMIM	MIM:604129
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL DOMINANT	OMIM	MIM:131750
COL7A1	EPIDERMOLYSIS BULLOSA DYSTROPHICA, AUTOSOMAL RECESSIVE	OMIM	MIM:226600
COL7A1	EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN [...]	OMIM	MIM:132000
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OMIM	MIM:131705

SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138	OMIM
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750	OMIM
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808	orphanet
TREX1	Aicardi-Goutieres Syndrome	OrphaNet	OrphaNet:51	orphanet
TREX1	HERNS syndrome	OrphaNet	OrphaNet:63261	orphanet
TREX1	CHILBLAIN LUPUS	OrphaNet	OrphaNet:90280	orphanet
TREX1	Cerebroretinal vasculopathy	OrphaNet	OrphaNet:3421	orphanet
TREX1	Hereditary vascular retinopathy	OrphaNet	OrphaNet:71291	orphanet
COL7A1	Generalized dominant dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:231568	orphanet
COL7A1	Dystrophic epidermolysis bullosa pruriginosa	OrphaNet	OrphaNet:89843	orphanet
COL7A1	Recessive dystrophic epidermolysis bullosa inversa	OrphaNet	OrphaNet:79409	orphanet
COL7A1	Dystrophic epidermolysis bullosa, nails only	OrphaNet	OrphaNet:158676	orphanet
COL7A1	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN	OrphaNet	OrphaNet:79411	orphanet
COL7A1	Centripetalis recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:89841	orphanet
COL7A1	Acral dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:158673	orphanet
COL7A1	Recessive dystrophic epidermolysis bullosa-generalized other	OrphaNet	OrphaNet:89842	orphanet
COL7A1	Epidermolysis bullosa simplex superficialis	OrphaNet	OrphaNet:89839	orphanet
COL7A1	Severe generalized recessive dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79408	orphanet
COL7A1	Pretibial dystrophic epidermolysis bullosa	OrphaNet	OrphaNet:79410	orphanet
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159	orphanet

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
CELSR3	missense variant	ENST00000164024	NM_001407.2	ENSP00000164024	2	2	tolerated, ?	benign, ?	2
IP6K2	missense variant	ENST00000417896	?	ENSP00000388116	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000433104	?	ENSP00000396904	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000443964	?	ENSP00000410950	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000449610	?	ENSP00000393077	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000453202	?	ENSP00000387394	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000446860	NM_001190317.1	ENSP00000399052	G/D	gGt/gAt	?	?	1
IP6K2	missense variant	ENST00000431721	NM_001190316.1	ENSP00000414139	G/D	gGt/gAt	?	?	1
SLC26A6	missense variant	ENST00000614797	?	ENSP00000483669	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000383733	NM_134426.2	ENSP00000373239	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000395550	NM_022911.2	ENSP00000378920	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000431739	?	ENSP00000401813	H/R	cAt/cGt	?	?	1
SLC26A6	missense variant	ENST00000444531	?	ENSP00000404607	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000455886	NM_001281732.1	ENSP00000401066	H/R	cAt/cGt	?	?	1
SLC26A6	missense variant	ENST00000421649	?	ENSP00000389922	M/V	Atg/Gtg	?	?	1
SLC26A6	missense variant	ENST00000358747	NM_001040454.1	ENSP00000351597	M/V	Atg/Gtg	?	?	1
SLC26A6	missense	ENST00000420764	NM_134263.2	ENSP00000404684	M/V	Atg/Gtg	?	?	1

	variant								
SLC26A6	missense variant	ENST00000414944	?	ENSP00000388721	H/R	cAt/cGt	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
WDR6	ENST00000615452	ILMN_1669484	skin	1.74×10-39 (p-value)	MuTHER consortium	19
WDR6	ENST00000452875		adipocyte	1.88×10-33 (p-value)	MuTHER consortium	19
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	skin	1.46×10-7 (p-value)	MuTHER consortium	19
USP4	ENST00000483212		adipocyte	5.95×10-12 (p-value)	MuTHER consortium	19
USP4	ENST00000485450		blood	7.52×10-5 (p-value)	MuTHER consortium	19
C3orf62	ENST00000343010					
TMEM89	ENST00000330862	ILMN_1694339	skin	2.13×10-7 (p-value)	MuTHER consortium	19
			adipocyte	3.59×10-5 (p-value)	MuTHER consortium	19
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	4.30×10-16 (p-value)	MuTHER consortium	19
RP11-3B7.1	ENST00000440528					
ATRIP	ENST00000320211	ILMN_2370464	skin	3.13×10-5 (p-value)	MuTHER consortium	1
TREX1	ENST00000436480					
ATRIP	ENST00000346691					
TREX1	ENST00000395677					
TREX1	ENST00000296443					
TREX1	ENST00000424906					
ATRIP	ENST00000412052					
TREX1	ENST00000433541					
TREX1	ENST00000629913					
TREX1	ENST00000357105					
TREX1	ENST00000625293					
WDR6	?	ENSG00000178252	lung	2.56×10-11 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	lung	5.04×10-8 (p-value)	GTEx Portal V6	75
CELSR3	?	ENSG00000008300	lung	2.09×10-5 (p-value)	GTEx Portal V6	2
WDR6	?	ENSG00000178252	transformed fibroblasts	7.13×10-18 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	transformed fibroblasts	5.36×10-9 (p-value)	GTEx Portal V6	73
CCDC36	?	ENSG00000173421	transformed fibroblasts	8.39×10-7 (p-value)	GTEx Portal V6	51
IP6K2	?	ENSG00000068745	transformed fibroblasts	1.54×10-7 (p-value)	GTEx Portal V6	65
RP11-3B7.1	?	ENSG00000225399	transformed fibroblasts	1.01×10-5 (p-value)	GTEx Portal V6	11
WDR6	?	ENSG00000178252	blood	8.00×10-12 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	blood	1.10×10-5 (p-value)	GTEx Portal V6	3
WDR6	?	ENSG00000178252	tibial artery	4.82×10-9 (p-value)	GTEx Portal V6	75
TMEM89	?	ENSG00000183396	tibial artery	1.56×10-8 (p-value)	GTEx Portal V6	74

IP6K2	?	ENSG00000068745	tibial artery	6.46×10-7 (p-value)	GTEx Portal V6	54
NICN1	?	ENSG00000145029	tibial artery	5.27×10-5 (p-value)	GTEx Portal V6	2
WDR6	ENST00000615452	ILMN_1669484	monocyte	2.40×10-14 (p-value)	Fairfax et al.	13
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000395474					
WDR6	ENST00000610967					
WDR6	ENST00000608424					
WDR6	ENST00000492780					
WDR6	ENST00000420783					
UBA7	ENST00000497908	ILMN_1794612	b-cell	6.47×10-5 (p-value)	Fairfax et al.	8
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
NCKIPSD	ENST00000294129	ILMN_1736623	monocyte	2.22×10-4 (p-value)	Fairfax et al.	4
NCKIPSD	ENST00000416649					
NCKIPSD	ENST00000470006					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.21×10-9 (p-value)	Fairfax et al.	13
USP4	ENST00000483212		b-cell	1.64×10-6 (p-value)	Fairfax et al.	13
C3orf62	ENST00000343010					
USP4	ENST00000485450					
SLC26A6	ENST00000420764	ILMN_1785252	b-cell	3.59×10-5 (p-value)	Fairfax et al.	6
SLC26A6	ENST00000395550					
SLC26A6	ENST00000489483					
SLC26A6	ENST00000480524					
SLC26A6	ENST00000455886					
SLC26A6	ENST00000307364					
SLC26A6	ENST00000614797					
SLC26A6	ENST00000358747					
SLC26A6	ENST00000383733					
SLC26A6	ENST00000337000					
RBM6	ENST00000442092	ILMN_1738239	monocyte	1.47×10-5 (p-value)	Fairfax et al.	7
RBM6	ENST00000266022		b-cell	2.57×10-5 (p-value)	Fairfax et al.	7
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
WDR6	?	ENSG00000178252	skeletal muscle	1.54×10-13 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	skeletal muscle	1.48×10-13 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	skeletal muscle	1.01×10-5 (p-value)	GTEx Portal V6	20
SLC26A6	?	ENSG00000225697	EBV lymphocytes	1.46×10-6 (p-value)	GTEx Portal V6	63
WDR6	?	ENSG00000178252	EBV lymphocytes	9.15×10-7 (p-value)	GTEx Portal V6	43
WDR6	?	ENSG00000178252	sun exposed skin	1.46×10-35 (p-value)	GTEx Portal V6	75
WDR6	?	ENSG00000178252	unexposed skin	1.57×10-22 (p-value)	GTEx Portal V6	75
TMEM89	?	ENSG00000183396	aorta	2.36×10-5 (p-value)	GTEx Portal V6	7
IP6K2	?	ENSG00000068745	aorta	1.01×10-5 (p-value)	GTEx Portal V6	25

WDR6	?	ENSG00000178252	subcutaneous adipocytes	2.01×10-21 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	subcutaneous adipocytes	3.25×10-5 (p-value)	GTEx Portal V6	12
NCKIPSD	?	ENSG00000213672	visceral adipocytes	2.83×10-7 (p-value)	GTEx Portal V6	71
WDR6	?	ENSG00000178252	visceral adipocytes	1.53×10-7 (p-value)	GTEx Portal V6	55
WDR6	?	ENSG00000178252	cortex	5.42×10-7 (p-value)	GTEx Portal V6	68
WDR6	?	ENSG00000178252	tibial nerve	7.26×10-13 (p-value)	GTEx Portal V6	75
SLC26A6	?	ENSG00000225697	tibial nerve	4.59×10-5 (p-value)	GTEx Portal V6	10
WDR6	?	ENSG00000178252	esophagus mucosa	3.87×10-26 (p-value)	GTEx Portal V6	75
NCKIPSD	?	ENSG00000213672	esophagus mucosa	1.58×10-8 (p-value)	GTEx Portal V6	74
PFKFB4	ENST00000232375	ILMN_1653292	blood	6.11×10-5 (p-value)	Westra et al.	9
CCDC36	?	ENSG00000173421	testis	4.13×10-7 (p-value)	GTEx Portal V6	54
CCDC36	?	ENSG00000173421	muscularis mucosae	2.23×10-6 (p-value)	GTEx Portal V6	57
KLHDC8B	?	ENSG00000185909	muscularis mucosae	8.00×10-6 (p-value)	GTEx Portal V6	24
WDR6	?	ENSG00000178252	blood	2.09×10-3 (q-value)	SeeQTL DB (HapMap)	13
WDR6	ENST00000615452	ILMN_1669484	monocyte	5.52×10-35 (p-value)	Zeller et al.	7
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
USP4	ENST00000351842	ILMN_1798712	monocyte	1.59×10-26 (p-value)	Zeller et al.	7
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
WDR6	?	ENSG00000178252	breast	3.91×10-6 (p-value)	GTEx Portal V6	25
SLC26A6	?	ENSG00000225697	thyroid	1.41×10-5 (p-value)	GTEx Portal V6	1
WDR6	?	ENSG00000178252	stomach	9.03×10-7 (p-value)	GTEx Portal V6	32
CCDC71	?	ENSG00000177352	cerebellar hemisphere	5.33×10-6 (p-value)	GTEx Portal V6	36
WDR6	?	ENSG00000178252	pancreas	3.37×10-5 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000338496	1	ENCP00000036385	SHISA5
		ENCP00000036431	ARIH2
		ENCP00000036356	ZNF589
		ENCP00000036451	DALRD3
		ENCP00000036473	QARS
		ENCP00000036376	CCDC51

		ENCP00000036448	WDR6
		ENCP00000036353	CDC25A
ENCE00000338642	1	ENCP00000036430	SLC25A20
		ENCP00000036466	QRICH1
		ENCP00000036373	PLXNB1
		ENCP00000036398	COL7A1 COL7A1
		ENCP00000036399	COL7A1 COL7A1
		ENCP00000036419	NCKIPSD
		ENCP00000036367	PLXNB1
		ENCP00000036396	COL7A1 COL7A1
ENCE00000338674	1	ENCP00000036410	SLC26A6
		ENCP00000036434	ARIH2
		ENCP00000036384	SHISA5
ENCE00000338847	1	ENCP00000036433	ARIH2
ENCE00000338585	1	ENCP00000036360	SPINK8
		ENCP00000036439	ARIH2
		ENCP00000036380	ATRIP
		ENCP00000036426	IP6K2
ENCE00000338754	1	ENCP00000036427	PRKAR2A
		ENCP00000036409	SLC26A6

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363494 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, Rad21, TAF7, Sin3Ak20, H3K9ac, CTCF, TAF1, H3K4me2, SP1, H3K4me3, H3K27ac
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H3K9ac, H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	DNase1, H3K4me2, H3K9ac, H3K27ac, CTCF, H3K4me3, H3K27me3, H3K36me3, H3K4me1, TAF1, PolII, H3K79me2, Rad21
		blood (GM12878)	PolII, H2AZ, DNase1, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		lung (IMR90)	CTCF, DNase1, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac
		NHLF	H3K27ac, H3K9ac, CTCF, DNase1, H3K4me3
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K4me2, H3K9ac, H3K36me3, H3K4me3, DNase1
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1
ENSR00001363497 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	H3K36me3
ENSR00001674778 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, PolII, Rad21, Egr1, H3K9ac, CTCF, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K27me3
		skin (NHDF-AD)	CTCF, H3K4me2
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, CTCF, H3K4me3
		lung (IMR90)	CTCF
		blood (GM12878)	DNase1, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K4me2
		skin (NHEK)	DNase1, CTCF, H3K4me3
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2

		breast (HMEC)	H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, CTCF, DNase1
		A549	H3K4me3, DNase1
ENSR00001674779 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K9ac, H3K4me3, H3K27me3, PolII, Jund, TAF7, p300, Nrsf, Sin3Ak20, Yy1, HDAC2, TAF1, H3K4me2, DNase1
		HSMMtube	H3K4me2, H2AZ, H3K27me3
		blood (K562)	DNase1, Nrsf, PolII, Sin3Ak20, Ini1, Max
		skin (NHDF-AD)	DNase1, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	Yy1, Nrsf, Sin3Ak20, PolII, TAF1, H3K9ac, H3K4me3, DNase1
		lung (IMR90)	H3K4me3, H3K27me3, H3K4me2
		blood (GM12878)	DNase1, Sin3Ak20, Nrsf, PolII, Yy1, H3K4me3
		nervous (NH-A)	H3K4me2
		skin (NHEK)	H3K9ac, H3K4me3, H3K27me3, DNase1
		NHLF	H3K27me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	Ini1, Nrsf, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, H3K27me3
		endothelium (HUVEC)	Max, H3K4me3, H3K27me3, DNase1
		A549	H3K4me3, H3K9ac, DNase1
ENSR00001478981 (promoter flanking region)	3	embryonic stem cell (H1ESC)	PolII, H3K9ac, H3K27ac, H3K4me2, H3K36me3, DNase1, H3K27me3
		HSMMtube	H3K79me2, H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K79me2, H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, H3K27ac, DNase1
		blood (GM12878)	PolII, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
		NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, H3K4me1, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001363512 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	H3K79me2, CTCF, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363515 (promoter flanking region)	1	NHLF	H3K4me1, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K4me2, H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K36me3, H3K79me2

		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K79me2, H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H4K20me1, H3K79me2, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K4me1, DNase1, H3K36me3, H3K4me2, H3K27ac
ENSR00001363517	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3
(promoter flanking region)		HSMMtube	DNase1, H3K79me2, H3K9ac, H3K4me2, H3K4me3, H3K36me3
		blood (K562)	HEY1, H2AZ, TAF1, ELF1, Cfos, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K79me2, H3K9ac, Yy1, H3K27ac, Max
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	DNase1, H3K36me3, H3K27me3, H3K4me3, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, DNase1, H3K79me2
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K36me3, H3K9ac, DNase1, H3K4me3
		Osteobl	H3K27me3, H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K27me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	DNase1, H3K36me3, H3K27me3, PolII, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC141002.1	upstream gene variant	1996	ENST00000619404	?	?	4
CELSR3	downstream gene variant, upstream gene variant	1483	ENST00000470999	?	?	6
CELSR3	downstream gene variant, upstream gene variant	1568	ENST00000164024	NM_001407.2	ENSP00000164024	8
CELSR3	downstream gene variant, upstream gene variant	1295	ENST00000498057	?	?	7
CELSR3	downstream gene variant, upstream gene variant	1295	ENST00000461362	?	?	7
CELSR3-AS1	upstream gene variant, downstream gene variant	1690	ENST00000421275	?	?	7
IP6K2	downstream gene variant	1429	ENST00000433104	?	ENSP00000396904	1
IP6K2	downstream gene variant, upstream gene variant	122	ENST00000424035	?	ENSP00000406357	6
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000443964	?	ENSP00000410950	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000446860	NM_001190317.1	ENSP00000399052	4
IP6K2	downstream gene variant, upstream gene variant	241	ENST00000416707	?	ENSP00000387759	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000454335	?	ENSP00000412121	4
IP6K2	downstream gene variant, upstream gene variant	1957	ENST00000450045	?	ENSP00000394488	4
IP6K2	downstream gene variant, upstream gene variant	256	ENST00000436134	?	?	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000443853	?	ENSP00000389761	4
IP6K2	downstream gene variant, upstream gene variant	252	ENST00000449610	?	ENSP00000393077	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	4
IP6K2	downstream gene variant, upstream gene variant	228	ENST00000431721	NM_001190316.1	ENSP00000414139	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000449563	?	ENSP00000411776	4
IP6K2	downstream gene variant, upstream gene	195	ENST00000412795	?	ENSP00000389755	4

	variant					
IP6K2	downstream gene variant, upstream gene variant	81	ENST00000440424	?	ENSP00000393797	6
IP6K2	downstream gene variant, upstream gene variant	278	ENST00000413654	?	ENSP00000414428	4
IP6K2	downstream gene variant, upstream gene variant	83	ENST00000434860	?	ENSP00000388420	6
IP6K2	downstream gene variant, upstream gene variant	166	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	4
IP6K2	downstream gene variant, upstream gene variant	229	ENST00000412850	?	ENSP00000395819	4
IP6K2	downstream gene variant, upstream gene variant	251	ENST00000476645	?	?	8
IP6K2	downstream gene variant, upstream gene variant	1915	ENST00000455545	?	ENSP00000410454	4
IP6K2	downstream gene variant, upstream gene variant	238	ENST00000417896	?	ENSP00000388116	4
IP6K2	downstream gene variant, upstream gene variant	230	ENST00000437427	?	ENSP00000403968	4
IP6K2	downstream gene variant, upstream gene variant	194	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	4
IP6K2	downstream gene variant, upstream gene variant	82	ENST00000453202	?	ENSP00000387394	5
IP6K2	downstream gene variant, upstream gene variant	1455	ENST00000491686	?	?	5
IP6K2	downstream gene variant, upstream gene variant	234	ENST00000413298	?	ENSP00000396203	4
IP6K2	downstream gene variant, upstream gene variant	486	ENST00000479914	?	?	4
MIR4793	downstream gene variant, upstream gene variant	574	ENST00000577502	?	?	4
MIR6824	downstream gene variant, upstream gene variant	242	ENST00000611478	?	?	14
NCKIPSD	downstream gene variant, upstream gene variant	671	ENST00000294129	NM_016453.3	ENSP00000294129	6
NCKIPSD	downstream gene variant, upstream gene variant	577	ENST00000453349	?	ENSP00000408588	7
NCKIPSD	downstream gene variant, upstream gene variant	573	ENST00000454134	?	ENSP00000416144	7
NCKIPSD	downstream gene variant, upstream gene variant	577	ENST00000426678	?	ENSP00000416904	7
NCKIPSD	downstream gene variant, upstream gene variant	236	ENST00000439518	?	ENSP00000409675	7
NCKIPSD	downstream gene variant, upstream gene variant	671	ENST00000416649	NM_184231.2	ENSP00000389059	6
NCKIPSD	downstream gene variant, upstream gene variant	1106	ENST00000470006	?	?	10
NCKIPSD	downstream gene variant, upstream gene variant	436	ENST00000415281	?	ENSP00000406442	8
NCKIPSD	downstream gene variant, upstream gene variant	1193	ENST00000413374	?	ENSP00000396683	10
RP11-572O6.1	upstream gene variant, downstream gene variant	359	ENST00000607025	?	?	12
SLC26A6	downstream gene variant, upstream gene variant	14	ENST00000431213	?	ENSP00000387532	9
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000337000	NM_001281733.1	ENSP00000337648	6
SLC26A6	downstream gene variant, upstream gene variant	722	ENST00000494717	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	973	ENST00000414944	?	ENSP00000388721	8
SLC26A6	downstream gene variant, upstream gene variant	614	ENST00000485361	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	1212	ENST00000469693	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	314	ENST00000482282	?	?	11
SLC26A6	downstream gene variant, upstream gene variant	962	ENST00000444531	?	ENSP00000404607	8
SLC26A6	downstream gene variant, upstream gene variant	5	ENST00000426599	?	ENSP00000405872	9
SLC26A6	downstream gene variant, upstream gene variant	425	ENST00000421649	?	ENSP00000389922	8

SLC26A6	downstream gene variant, upstream gene variant	977	ENST00000431739	?	ENSP00000401813	8
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000395550	NM_022911.2	ENSP00000378920	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000383733	NM_134426.2	ENSP00000373239	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000358747	NM_001040454.1	ENSP00000351597	9
SLC26A6	downstream gene variant, upstream gene variant	578	ENST00000466257	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000307364	?	ENSP00000307089	6
SLC26A6	downstream gene variant, upstream gene variant	112	ENST00000614797	?	ENSP00000483669	6
SLC26A6	downstream gene variant, upstream gene variant	133	ENST00000455886	NM_001281732.1	ENSP00000401066	6
SLC26A6	downstream gene variant, upstream gene variant	74	ENST00000489483	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	114	ENST00000480524	?	?	9
SLC26A6	downstream gene variant, upstream gene variant	667	ENST00000496469	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	1206	ENST00000462009	?	?	8
SLC26A6	downstream gene variant, upstream gene variant	110	ENST00000420764	NM_134263.2	ENSP00000404684	6
TMEM89	upstream gene variant	2697	ENST00000330862	NM_001008269.1	ENSP00000329557	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	3	3	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SLC26A6	ENST00000358747	NM_001040454.1	ENSP00000351597	1
SLC26A6	ENST00000395550	NM_022911.2	ENSP00000378920	1
SLC26A6	ENST00000431739	?	ENSP00000401813	1
SLC26A6	ENST00000489483	?	?	1
SLC26A6	ENST00000426599	?	ENSP00000405872	1
SLC26A6	ENST00000482282	?	?	1
SLC26A6	ENST00000455886	NM_001281732.1	ENSP00000401066	1
SLC26A6	ENST00000337000	NM_001281733.1	ENSP00000337648	1
SLC26A6	ENST00000614797	?	ENSP00000483669	1
SLC26A6	ENST00000307364	?	ENSP00000307089	1
SLC26A6	ENST00000480524	?	?	1
SLC26A6	ENST00000383733	NM_134426.2	ENSP00000373239	1
SLC26A6	ENST00000420764	NM_134263.2	ENSP00000404684	1

5'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
NCKIPSD	ENST00000426678	?	ENSP00000416904	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	11
CELSR3	ENST00000470999	?	?	2
CELSR3	ENST00000498057	?	?	5
CELSR3	ENST00000461362	?	?	5
CELSR3-AS1	ENST00000421275	?	?	3
IP6K2	ENST00000449563	?	ENSP00000411776	18
IP6K2	ENST00000455545	?	ENSP00000410454	18
IP6K2	ENST00000436134	?	?	18
IP6K2	ENST00000440424	?	ENSP00000393797	16
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	17
IP6K2	ENST00000454335	?	ENSP00000412121	18
IP6K2	ENST00000433104	?	ENSP00000396904	23
IP6K2	ENST00000413654	?	ENSP00000414428	18
IP6K2	ENST00000443964	?	ENSP00000410950	17
IP6K2	ENST00000432678	NM_001146178.2, NM_001146179.2	ENSP00000400812	18
IP6K2	ENST00000416707	?	ENSP00000387759	18
IP6K2	ENST00000449610	?	ENSP00000393077	17
IP6K2	ENST00000450045	?	ENSP00000394488	18
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	17
IP6K2	ENST00000434860	?	ENSP00000388420	16
IP6K2	ENST00000437427	?	ENSP00000403968	18
IP6K2	ENST00000340879	NM_001005910.2, NM_001005911.2	ENSP00000341925	18
IP6K2	ENST00000424035	?	ENSP00000406357	16
IP6K2	ENST00000417896	?	ENSP00000388116	17
IP6K2	ENST00000476645	?	?	13
IP6K2	ENST00000443853	?	ENSP00000389761	18
IP6K2	ENST00000412795	?	ENSP00000389755	18
IP6K2	ENST00000413298	?	ENSP00000396203	18
IP6K2	ENST00000491686	?	?	1
IP6K2	ENST00000328631	NM_001005909.2, NM_016291.3	ENSP00000331103	19
IP6K2	ENST00000412850	?	ENSP00000395819	18
IP6K2	ENST00000479914	?	?	3
NCKIPSD	ENST00000426678	?	ENSP00000416904	2
NCKIPSD	ENST00000470006	?	?	6
NCKIPSD	ENST00000413374	?	ENSP00000396683	6
NCKIPSD	ENST00000453349	?	ENSP00000408588	2
NCKIPSD	ENST00000416649	NM_184231.2	ENSP00000389059	11
NCKIPSD	ENST00000294129	NM_016453.3	ENSP00000294129	11
NCKIPSD	ENST00000454134	?	ENSP00000416144	2
NCKIPSD	ENST00000439518	?	ENSP00000409675	2
NCKIPSD	ENST00000415281	?	ENSP00000406442	3

SLC26A6	ENST00000358747	NM_001040454.1	ENSP00000351597	5
SLC26A6	ENST00000395550	NM_022911.2	ENSP00000378920	8
SLC26A6	ENST00000431739	?	ENSP00000401813	4
SLC26A6	ENST00000489483	?	?	7
SLC26A6	ENST00000426599	?	ENSP00000405872	4
SLC26A6	ENST00000469693	?	?	3
SLC26A6	ENST00000482282	?	?	2
SLC26A6	ENST00000455886	NM_001281732.1	ENSP00000401066	8
SLC26A6	ENST00000431213	?	ENSP00000387532	2
SLC26A6	ENST00000462009	?	?	3
SLC26A6	ENST00000337000	NM_001281733.1	ENSP00000337648	9
SLC26A6	ENST00000614797	?	ENSP00000483669	8
SLC26A6	ENST00000444531	?	ENSP00000404607	2
SLC26A6	ENST00000307364	?	ENSP00000307089	9
SLC26A6	ENST00000414944	?	ENSP00000388721	2
SLC26A6	ENST00000480524	?	?	6
SLC26A6	ENST00000421649	?	ENSP00000389922	2
SLC26A6	ENST00000496469	?	?	4
SLC26A6	ENST00000383733	NM_134426.2	ENSP00000373239	8
SLC26A6	ENST00000420764	NM_134263.2	ENSP00000404684	8

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CELSR3	ENST00000164024	NM_001407.2	ENSP00000164024	1
IP6K2	ENST00000446860	NM_001190317.1	ENSP00000399052	1
IP6K2	ENST00000433104	?	ENSP00000396904	1
IP6K2	ENST00000443964	?	ENSP00000410950	1
IP6K2	ENST00000416707	?	ENSP00000387759	1
IP6K2	ENST00000449610	?	ENSP00000393077	1
IP6K2	ENST00000431721	NM_001190316.1	ENSP00000414139	1
IP6K2	ENST00000417896	?	ENSP00000388116	1
IP6K2	ENST00000412795	?	ENSP00000389755	1
SLC26A6	ENST00000431213	?	ENSP00000387532	1
SLC26A6	ENST00000307364	?	ENSP00000307089	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IP6K2	ENST00000433104	?	ENSP00000396904	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CELSR3	ENST00000498057	?	3
CELSR3	ENST00000461362	?	3
IP6K2	ENST00000436134	?	1

SLC26A6	ENST00000489483	?	4
SLC26A6	ENST00000482282	?	2
SLC26A6	ENST00000485361	?	1
SLC26A6	ENST00000494717	?	1
SLC26A6	ENST00000480524	?	3