Locus 227 (rs7184253)

SNiPAcard

Block annotations

Block info

genomic range	chr16:67,308,279-67,549,746
block size	241,468 bp
variant count	114 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.038 [-3.819 – 3.611]	gene(s) hit or close-by	AGRP, ATP6V0D1, CTD-2012K14.8, FAM65A, HSD11B2, KCTD19, LRRC36, PLEKHG4, RN7SKP118, RNU1-123P, RP11-297D21.2, RP11-297D21.4, SLC9A5, TPPP3, ZDHHC1
phastCons	μ = 0.206 [0 – 1]	eQTL gene(s)	AGRP, ATP6V0D1, C16orf86, CTCF, DPEP2, DUS2, EXOC3L1, HSD11B2, LCAT, LRRC36, NFATC3, PLA2G15, RANBP10, TRADD, ZDHHC1
GERP++	μ = 0.189 [-11.3 – 5.69]	potentially regulated gene(s)	ATP6V0D1, C16orf70, C16orf86, CBFB, CES4A, EDC4, ELMO3, EXOC3L1, FAM65A, FBXL8, GFOD2, HSD11B2, HSF4, LCAT, NUTF2, SLC12A4, TPPP3, TRADD, ZDHHC1
CADD score	μ = 6.295 [0.006 – 32]	disease gene(s)	PLEKHG4, HSD11B2, LCAT, HSF4, CTCF, CBFB, AGRP

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Leanness, inherited	association	ClinVar	RCV000007754.2	1
Obesity, late-onset	association	ClinVar	RCV000007753.1	1
?	HGMD curated	HGMD	CM057710	1
?	HGMD curated	HGMD	CM023599	1
OBESITY, LATE-ONSET	OMIM curated	OMIM	MIM:602311	1

Disease gene annotation

gene	trait	source DB	source entry/link
PLEKHG4	Spinocerebellar ataxia type 4	OrphaNet	OrphaNet:98765
HSD11B2	APPARENT MINERALOCORTICOID EXCESS	OrphaNet	OrphaNet:320
LCAT	FISH-EYE DISEASE	OrphaNet	OrphaNet:79292
LCAT	Familial LCAT deficiency	OrphaNet	OrphaNet:79293
HSF4	Zonular cataract	OrphaNet	OrphaNet:98995
CTCF	Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome	OrphaNet	OrphaNet:363611
CBFB	Acute myeloid leukemia with abnormal bone marrow eosinophils inv(16)(p13q22) or t(16;16)(p13;q22)	OrphaNet	OrphaNet:98829
HSD11B2	APPARENT MINERALOCORTICOID EXCESS	OMIM	MIM:218030
AGRP	OBESITYLEANNESS, INCLUDED	OMIM	MIM:601665
LCAT	FISH-EYE DISEASE	OMIM	MIM:136120
LCAT	LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY	OMIM	MIM:245900
HSF4	CATARACT 5, MULTIPLE TYPES	OMIM	MIM:116800
CTCF	MENTAL RETARDATION, AUTOSOMAL DOMINANT 21	OMIM	MIM:615502
HSF4	cataract Marner type (CAM)	DECIPHER	MIM:116800
HSF4	cataract zonular HSF4-related (CZ-HSF4)	DECIPHER	MIM:116800

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)
AGRP	missense variant	ENST00000290953	NM_001138.1	ENSP00000290953	T/A	Act/Gct	?	?	1
KCTD19	missense variant	ENST00000304372	NM_001100915.1	ENSP00000305702	K/E	Aag/Gag	?	?	1
LRRC36	missense variant	ENST00000435835	?	ENSP00000411122	2	2	tolerated, ?	benign, ?	2
LRRC36	missense variant	ENST00000565019	?	ENSP00000464675	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000563189	NM_001161575.1	ENSP00000455103	2	3	?, tolerated	benign, ?	3
LRRC36	missense variant	ENST00000569499	?	ENSP00000456814	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000329956	NM_018296.5	ENSP00000329943	2	3	?, tolerated	benign, ?	3
LRRC36	missense variant	ENST00000568010	?	ENSP00000455018	R/P	cGt/cCt	?	benign	1
LRRC36	missense variant	ENST00000561948	?	ENSP00000456347	R/P	cGt/cCt	?	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LRRC36	?	ENSG00000159708	skeletal muscle	3.14×10^{-17} (p-value)	GTEx Portal V6	111
C16orf86	?	ENSG00000159761	skeletal muscle	1.11×10^{-5} (p-value)	GTEx Portal V6	24
RANBP10	?	ENSG00000141084	cortex	2.02×10^{-6} (p-value)	GTEx Portal V6	14
RANBP10	?	ENSG00000141084	tibial nerve	6.59×10^{-11} (p-value)	GTEx Portal V6	111
LRRC36	?	ENSG00000159708	thyroid	3.22×10^{-15} (p-value)	GTEx Portal V6	111
ZDHHC1	?	ENSG00000159714	thyroid	1.15×10^{-7} (p-value)	GTEx Portal V6	105
HSD11B2	?	ENSG00000176387	thyroid	6.59×10^{-17} (p-value)	GTEx Portal V6	110
DUS2	?	ENSG00000167264	thyroid	1.69×10^{-5} (p-value)	GTEx Portal V6	10
RANBP10	?	ENSG00000141084	thyroid	3.72×10^{-5} (p-value)	GTEx Portal V6	1
EXOC3L1	?	ENSG00000179044	thyroid	1.60×10^{-5} (p-value)	GTEx Portal V6	1
LRRC36	?	ENSG00000159708	visceral adipocytes	5.09×10^{-9} (p-value)	GTEx Portal V6	55
LRRC36	?	ENSG00000159708	atrial appendage	3.59×10^{-6} (p-value)	GTEx Portal V6	21
RANBP10	?	ENSG00000141084	tibial artery	1.57×10^{-7} (p-value)	GTEx Portal V6	83
CTCF	?	ENSG00000102974	tibial artery	1.06×10^{-5} (p-value)	GTEx Portal V6	10
EXOC3L1	?	ENSG00000179044	tibial artery	1.61×10^{-5} (p-value)	GTEx Portal V6	3
DPEP2	?	ENSG00000167261	blood	2.93×10^{-8} (p-value)	GTEx Portal V6	22
PLA2G15	?	ENSG00000103066	blood	1.70×10^{-6} (p-value)	GTEx Portal V6	14
DUS2	?	ENSG00000167264	blood	9.27×10^{-9} (p-value)	GTEx Portal V6	36
ATP6V0D1	?	ENSG00000159720	blood	1.22×10^{-15} (p-value)	GTEx Portal V6	58
ZDHHC1	?	ENSG00000159714	transformed fibroblasts	1.65×10^{-5} (p-value)	GTEx Portal V6	11
DUS2	ENST00000432752	ILMN_1811650	monocyte	4.02×10^{-9} (p-value)	Fairfax et al.	10

DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000358896					
DUS2	ENST00000565980					
ATP6V0D1	ENST00000568620	ILMN_1795826	monocyte	2.88×10-15 (p-value)	Fairfax et al.	10
ATP6V0D1	ENST00000567694		b-cell	6.99×10-8 (p-value)	Fairfax et al.	10
ATP6V0D1	ENST00000563064					
ATP6V0D1	ENST00000561852					
ATP6V0D1	ENST00000602876					
ATP6V0D1	ENST00000290949					
ATP6V0D1	ENST00000567170					
ATP6V0D1	ENST00000564615					
ATP6V0D1	ENST00000426604					
ATP6V0D1	ENST00000565835					
ATP6V0D1	ENST00000563305					
AGRP	ENST00000290953	ILMN_1714253	monocyte	4.43×10-10 (p-value)	Fairfax et al.	10
DUS2	ENST00000432752	ILMN_1811650	monocyte	4.71×10-45 (p-value)	Zeller et al.	5
DUS2	ENST00000565263					
DUS2	ENST00000561965					
DUS2	ENST00000565980					
DUS2	ENST00000358896					
ZDHHC1	ENST00000565726	ILMN_2094313	monocyte	1.76×10-13 (p-value)	Zeller et al.	3
ZDHHC1	ENST00000566075					
ZDHHC1	ENST00000348579					
TRADD	ENST00000486556	ILMN_1765851	monocyte	8.13×10-26 (p-value)	Zeller et al.	5
TRADD	ENST00000566247					
TRADD	ENST00000563348					
TRADD	ENST00000345057					
C16orf86	ENST00000403458	ILMN_1697800	monocyte	2.94×10-14 (p-value)	Zeller et al.	5
C16orf86	ENST00000459925					
C16orf86	ENST00000602974					
C16orf86	ENST00000602365					
C16orf86	ENST00000445068					
RANBP10	?	ENSG00000141084	subcutaneous adipocytes	4.19×10-8 (p-value)	GTEx Portal V6	35
LRRC36	?	ENSG00000159708	subcutaneous adipocytes	9.09×10-7 (p-value)	GTEx Portal V6	35
RANBP10	?	ENSG00000141084	muscularis mucosae	1.09×10-5 (p-value)	GTEx Portal V6	50
LRRC36	?	ENSG00000159708	muscularis mucosae	7.71×10-7 (p-value)	GTEx Portal V6	32
LRRC36	?	ENSG00000159708	pancreas	3.84×10-15 (p-value)	GTEx Portal V6	35
LRRC36	?	ENSG00000159708	breast	1.39×10-7 (p-value)	GTEx Portal V6	35
ATP6V0D1	?	ENSG00000159720	EBV lymphocytes	6.90×10-7 (p-value)	GTEx Portal V6	35
ATP6V0D1	?	ENSG00000159720	spleen	9.21×10-11 (p-value)	GTEx Portal V6	34
LCAT	ENST00000264005	ILMN_1815102	skin	3.97×10-6 (p-value)	MuTHER consortium	18
LCAT	ENST00000573538					
LCAT	ENST00000570980					
LCAT	ENST00000570369					
AGRP	ENST00000290953	ILMN_1714253	blood	1.29×10-5 (p-value)	MuTHER consortium	17
TRADD	ENST00000486556	ILMN_1765851	skin	4.86×10-8 (p-value)	MuTHER consortium	18

TRADD	ENST00000566247		blood	5.33×10-11 (p-value)	MuTHER consortium	18
TRADD	ENST00000563348		adipocyte	3.93×10-18 (p-value)	MuTHER consortium	18
TRADD	ENST00000345057					
C16orf86	ENST00000403458	ILMN_1697800	blood	2.18×10-8 (p-value)	MuTHER consortium	18
C16orf86	ENST00000459925					
C16orf86	ENST00000602974					
C16orf86	ENST00000602365					
C16orf86	ENST00000445068					
NFATC3	ENST00000346183	ILMN_1800976	skin	1.55×10-5 (p-value)	MuTHER consortium	9
NFATC3	ENST00000329524					
NFATC3	ENST00000539828					
NFATC3	ENST00000566301					
NFATC3	ENST00000379165					
NFATC3	ENST00000563796					
NFATC3	ENST00000570212					
NFATC3	ENST00000563288					
NFATC3	ENST00000549350					
NFATC3	ENST00000349223					
NFATC3	ENST00000563319					
NFATC3	ENST00000575270					
NFATC3	ENST00000569766					
NFATC3	ENST00000567152					
NFATC3	ENST00000553077					
NFATC3	ENST00000562926					
NFATC3	ENST00000535127					
NFATC3	ENST00000346183	ILMN_2360028	skin	4.68×10-6 (p-value)	MuTHER consortium	8
NFATC3	ENST00000329524					
NFATC3	ENST00000539828					
NFATC3	ENST00000379165					
NFATC3	ENST00000566301					
NFATC3	ENST00000563796					
NFATC3	ENST00000570212					
NFATC3	ENST00000563288					
NFATC3	ENST00000549350					
NFATC3	ENST00000575270					
NFATC3	ENST00000349223					
NFATC3	ENST00000563319					
NFATC3	ENST00000569766					
NFATC3	ENST00000567152					
NFATC3	ENST00000553077					
NFATC3	ENST00000562926					
NFATC3	ENST00000535127					
ATP6V0D1	?	ENSG00000159720	esophagus mucosa	6.60×10-6 (p-value)	GTEx Portal V6	2
ZDHHC1	?	ENSG00000159714	testis	3.79×10-6 (p-value)	GTEx Portal V6	6
AGRP	?	ENSG00000159723	blood	2.95×10-3 (q-value)	SeeQTL DB (HapMap)	1

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ATP6V0D1	ENST00000568620	ILMN_1795826	chr16	monocyte	2.82×10-15 (p-value)	Zeller et al.	5
ATP6V0D1	ENST00000563064						
ATP6V0D1	ENST00000567694						
ATP6V0D1	ENST00000602876						

ATP6V0D1	ENST00000561852						
ATP6V0D1	ENST00000290949						
ATP6V0D1	ENST00000567170						
ATP6V0D1	ENST00000564615						
ATP6V0D1	ENST00000565835						
ATP6V0D1	ENST00000426604						
ATP6V0D1	ENST00000563305						
AGRP	ENST00000290953	ILMN_1714253	chr16	monocyte	5.25×10^{-30} (p-value)	Zeller et al.	5

Putative effect on regulation

Transcription factor binding site variation

transcription factor	binding motif	motif position	highly informative position	score change	variant(s)
USF1	MA0281.1	5	yes	-0.163	1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000192385	1	ENCP00000021593	EXOC3L1
		ENCP00000021592	HSF4
		ENCP00000021577	CES4A
ENCE00000192543	1	ENCP00000021598	TPPP3
ENCE00000192544	1	ENCP00000021598	TPPP3
		ENCP00000021601	ATP6V0D1
ENCE00000192549	1	ENCP00000021614	EDC4
		ENCP00000021601	ATP6V0D1
		ENCP00000021598	TPPP3
ENCE00000192432	1	ENCP00000021581	C16orf70
		ENCP00000021594	ELMO3
		ENCP00000021586	HSF4 FBXL8
		ENCP00000021613	NUTF2
		ENCP00000021599	ZDHHC1
		ENCP00000021603	FAM65A
		ENCP00000021618	LCAT
		ENCP00000021587	HSF4
		ENCP00000021600	HSD11B2
		ENCP00000021580	CBFB
		ENCP00000021588	HSF4
ENCE00000192693	1	ENCP00000021614	EDC4
ENCE00000192435	3	ENCP00000021609	GFOD2
		ENCP00000021581	C16orf70
		ENCP00000021613	NUTF2
		ENCP00000021608	C16orf86
		ENCP00000021618	LCAT
		ENCP00000021603	FAM65A
		ENCP00000021580	CBFB
		ENCP00000021619	SLC12A4
		ENCP00000021583	TRADD FBXL8
		ENCP00000021588	HSF4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001634343 (enhancer)	2	liver (HepG2)	H3K4me1
		HSMMtube	H3K27me3
ENSR00000508958 (promoter flanking region)	1	cervix (HeLa-S3)	H3K27ac, H3K4me1, DNase1
		endothelium (HUVEC)	Cjun
		HSMMtube	H3K27me3
ENSR00000508959 (CTCF binding site)	1	embryonic stem cell (H1ESC)	USF1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		muscle (HSMM)	H3K27me3
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	USF1, H3K4me1, H3K27ac, CTCF, DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1, H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001634345 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001634346 (enhancer)	1	endothelium (HUVEC)	Cjun
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
ENSR00001634348 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00001634351 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, DNase1, H3K4me1
		HSMMtube	DNase1
		Osteobl	H3K27me3
		blood (K562)	DNase1, H3K4me1
		breast (HMEC)	H3K4me1, H3K4me2
		muscle (HSMM)	H3K4me1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, H3K9ac, DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K27me3
		skin (NHEK)	H3K4me1
ENSR00000508965 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF, Rad21, Sin3Ak20, H3K4me2, H3K27me3, H3K4me1, DNase1
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K27me3
		blood (K562)	Egr1, H3K4me1, DNase1
		skin (NHDF-AD)	DNase1
		breast (HMEC)	CTCF, H3K4me2, H3K4me1, DNase1
		muscle (HSMM)	H3K4me1, H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K27me3, DNase1
		liver (HepG2)	CTCF, H3K4me1, H3K4me2, H3K27me3, DNase1
		blood (GM12878)	H3K27me3
		A549	DNase1
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, CTCF, DNase1
ENSR00001634352 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, CTCF, Rad21, DNase1
		HSMMtube	DNase1
		liver (HepG2)	CTCF, H3K4me1
		skin (NHEK)	CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	H3K4me1
ENSR00000508968	3	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21

(promoter flanking region)		HSMMtube	H3K4me1, H3K27ac, DNase1
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K4me1, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me1, H2AZ, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	CTCF, H3K27me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K9ac, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1
		NHLF	H3K27ac, DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me1, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K36me3
ENSR00000272763	1	NHLF	H3K27ac
(CTCF binding site)		embryonic stem cell (H1ESC)	Rad21, CTCF
		Osteobl	H3K27ac
		blood (K562)	H3K27me3, CTCF
		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
		breast (HMEC)	H3K4me1
		muscle (HSMM)	H3K4me1, H3K27ac, H3K36me3
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	CTCF
		skin (NHEK)	H3K4me1
ENSR00000508970	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
(enhancer)		Osteobl	H3K36me3
		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001634357	2	embryonic stem cell (H1ESC)	H4K5ac, H3K27ac, H3K27me3, H3K4me1, H3K4me2
(open chromatin region)		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3
		A549	H3K27me3
		skin (NHEK)	H3K27me3
ENSR00000052856	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21, H3K27me3
(CTCF binding site)		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF, DNase1
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K4me2, H3K9ac, CTCF, H3K27me3, DNase1
		blood (GM12878)	DNase1, Rad21, CTCF
		lung (IMR90)	H3K27me3, CTCF
		nervous (NH-A)	CTCF, H3K27me3
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF, H3K27me3
		Osteobl	CTCF, H3K27me3
		blood (DND-41)	H3K27me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF, H3K27me3, DNase1
		A549	CTCF
ENSR00001506017	2	embryonic stem cell (H1ESC)	CTCF, Rad21, H3K36me3
(CTCF binding site)		blood (DND-41)	H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3

		blood (GM12878)	PolII, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001634363 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	CTCF, H3K36me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	HNF4G, RXRA, H3K4me1, H3K4me2, HNF4A, H3K9ac, CTCF, H3K27ac, Cmyc, H3K36me3, DNase1
		lung (IMR90)	H3K36me3, H3K27ac
		blood (GM12878)	DNase1, H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
ENSR00001634364 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, DNase1
		blood (K562)	CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, H3K27ac, CTCF, DNase1
		blood (GM12878)	DNase1, Yy1, CTCF, H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	DNase1, H3K36me3
		A549	CTCF, H3K36me3
ENSR00000508978 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, DNase1
		HSMMtube	DNase1
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K36me3, DNase1, H3K27ac, H3K4me1, Rad21
		lung (IMR90)	H3K27ac, H3K36me3, CTCF
		blood (GM12878)	DNase1, Yy1, Rad21, CTCF, H3K36me3
		skin (NHEK)	CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1
		A549	CTCF, H3K36me3
ENSR00001634366 (promoter flanking region)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	CTCF, DNase1
		HSMMtube	DNase1
		Osteobl	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	CTCF
ENSR00000052861 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Jund, CTCF, DNase1
		HSMMtube	CTCF, DNase1

		blood (K562)	FOSL1, Rad21, CTCFL, H4K20me1, CTCF, DNase1
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	H3K79me2, CTCF, DNase1
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, CTCF, DNase1
		lung (IMR90)	H3K79me1, H3K36me3, CTCF
		blood (GM12878)	DNase1, Rad21, H3K79me2, CTCF
		nervous (NH-A)	H4K20me1, CTCF
		skin (NHEK)	CTCF, H3K36me3, DNase1
		NHLF	CTCF
		Osteobl	H4K20me1, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1, Jund
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1, DNase1, CTCF
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF, DNase1
		A549	CTCF
ENSR00001634368 (enhancer)	1	embryonic stem cell (H1ESC)	Rad21, Jund, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	FOSL1, Rad21, CTCFL, H4K20me1, CTCF, DNase1
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	H3K79me2, CTCF, DNase1
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, CTCF, DNase1
		lung (IMR90)	H3K79me1, CTCF
		blood (GM12878)	CTCF, DNase1, Rad21, H3K79me2
		nervous (NH-A)	H4K20me1, CTCF
		skin (NHEK)	DNase1, CTCF, H3K36me3
		NHLF	CTCF
		Osteobl	H4K20me1, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Jund, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF, DNase1
		A549	CTCF
ENSR00000508981 (enhancer)	1	monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001634370 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K27me3
ENSR00001506024 (promoter flanking region)	2	cervix (HeLa-S3)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
		embryonic stem cell (H1ESC)	H3K27me3
		blood (DND-41)	H3K36me3
		blood (K562)	Max, USF1, DNase1
		skin (NHDF-AD)	DNase1
ENSR00001634371 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27ac, H3K4me1
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
ENSR00000508990 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCFL, CTCF
		skin (NHDF-AD)	DNase1, CTCF
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF
		blood (GM12878)	BCLAF1, H3K4me1, DNase1, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, H3K27ac, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF

		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1
ENSR00001634373	3	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
(promoter flanking region)		HSMMtube	CTCF
		blood (K562)	DNase1, Rad21, CTCFL, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		muscle (HSMM)	DNase1, CTCF
		liver (HepG2)	Rad21, CTCF, H3K27me3
		blood (GM12878)	BCLAF1, H3K4me1, DNase1, Rad21, H3K27ac, H3K4me2, CTCF
		lung (IMR90)	CTCF, DNase1
		nervous (NH-A)	DNase1, CTCF, H3K4me2
		skin (NHEK)	DNase1, CTCF
		NHLF	CTCF
		Osteobl	H3K27ac, H3K4me2, CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	Jund, H3K27ac, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, Cjun, CTCF
		A549	CTCF, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
ATP6V0D1	1	ENST00000290949 ENST00000563305 ENST00000565835 ENST00000568620 ENST00000602876	hsa-miR-132-3p hsa-miR-212-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AGRP	downstream gene variant, upstream gene variant	1737	ENST00000290953	NM_001138.1	ENSP00000290953	6
ATP6V0D1	downstream gene variant, upstream gene variant	457	ENST00000561852	?	ENSP00000457744	8
ATP6V0D1	downstream gene variant, upstream gene variant	470	ENST00000563064	?	ENSP00000457105	8
ATP6V0D1	downstream gene variant, upstream gene variant	124	ENST00000540149	?	ENSP00000441282	8
ATP6V0D1	downstream gene variant, upstream gene variant	493	ENST00000567170	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	1885	ENST00000568298	?	?	6
ATP6V0D1	downstream gene variant	61	ENST00000426604	?	ENSP00000393910	4
ATP6V0D1	downstream gene variant, upstream gene variant	1306	ENST00000561658	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	2025	ENST00000563305	?	?	3
ATP6V0D1	downstream gene variant	431	ENST00000568101	?	ENSP00000457104	2
ATP6V0D1	downstream gene variant, upstream gene variant	473	ENST00000564615	?	ENSP00000455802	6
ATP6V0D1	downstream gene variant, upstream gene variant	268	ENST00000565835	?	ENSP00000463328	7
ATP6V0D1	downstream gene variant, upstream gene variant	756	ENST00000564788	?	ENSP00000455695	8
ATP6V0D1	downstream gene variant	701	ENST00000566322	?	ENSP00000455446	4
ATP6V0D1	downstream gene variant, upstream gene variant	1932	ENST00000602876	?	ENSP00000473515	6
ATP6V0D1	downstream gene variant, upstream gene variant	189	ENST00000567694	?	?	7
ATP6V0D1	downstream gene variant, upstream gene variant	1805	ENST00000290949	NM_004691.4	ENSP00000290949	6
ATP6V0D1	downstream gene variant, upstream gene variant	937	ENST00000568620	?	?	5
ATP6V0D1	downstream gene variant, upstream gene variant	736	ENST00000564191	?	ENSP00000456695	8

CTD-2012K14.8	downstream gene variant	2019	ENST00000613438	?	?	3
CTD-2012K14.8	downstream gene variant	2107	ENST00000621378	?	?	3
FAM65A	upstream gene variant	2575	ENST00000562116	?	ENSP00000455239	3
HSD11B2	upstream gene variant, downstream gene variant	1919	ENST00000566606	?	ENSP00000473429	4
HSD11B2	upstream gene variant, downstream gene variant	500	ENST00000326152	NM_000196.3	ENSP00000316786	4
HSD11B2	upstream gene variant, downstream gene variant	1792	ENST00000567684	?	?	4
HSD11B2	upstream gene variant, downstream gene variant	2646	ENST00000569303	?	?	5
KCTD19	downstream gene variant, upstream gene variant	1234	ENST00000566295	?	ENSP00000462732	6
KCTD19	downstream gene variant, upstream gene variant	1040	ENST00000567976	?	ENSP00000458045	4
KCTD19	downstream gene variant, upstream gene variant	592	ENST00000561625	?	?	7
KCTD19	downstream gene variant, upstream gene variant	602	ENST00000569333	?	?	7
KCTD19	downstream gene variant, upstream gene variant	568	ENST00000304372	NM_001100915.1	ENSP00000305702	7
KCTD19	downstream gene variant	3350	ENST00000562841	?	ENSP00000463249	1
KCTD19	downstream gene variant, upstream gene variant	617	ENST00000562721	?	ENSP00000462122	7
KCTD19	downstream gene variant, upstream gene variant	578	ENST00000566392	?	?	7
KCTD19	upstream gene variant	2160	ENST00000562860	?	?	3
KCTD19	downstream gene variant, upstream gene variant	605	ENST00000570049	?	?	7
KCTD19	downstream gene variant, upstream gene variant	571	ENST00000568736	?	?	7
LRRC36	upstream gene variant, downstream gene variant	2848	ENST00000569228	?	?	4
LRRC36	upstream gene variant, downstream gene variant	1510	ENST00000566558	?	?	7
LRRC36	upstream gene variant, downstream gene variant	756	ENST00000435835	?	ENSP00000411122	3
LRRC36	upstream gene variant, downstream gene variant	1155	ENST00000561948	?	ENSP00000456347	5
LRRC36	upstream gene variant, downstream gene variant	464	ENST00000569552	?	ENSP00000454309	2
LRRC36	upstream gene variant, downstream gene variant	1038	ENST00000329956	NM_018296.5	ENSP00000329943	5
LRRC36	upstream gene variant, downstream gene variant	1157	ENST00000570075	?	ENSP00000454794	5
LRRC36	upstream gene variant, downstream gene variant	729	ENST00000563189	NM_001161575.1	ENSP00000455103	3
LRRC36	upstream gene variant, downstream gene variant	1035	ENST00000565019	?	ENSP00000464675	5
LRRC36	upstream gene variant, downstream gene variant	741	ENST00000567823	?	ENSP00000456164	3
LRRC36	upstream gene variant, downstream gene variant	729	ENST00000567723	?	ENSP00000455799	3
LRRC36	upstream gene variant, downstream gene variant	638	ENST00000561821	?	ENSP00000456517	2
LRRC36	upstream gene variant, downstream gene variant	276	ENST00000569121	?	?	6
LRRC36	upstream gene variant, downstream gene variant	721	ENST00000568010	?	ENSP00000455018	3
LRRC36	upstream gene variant, downstream gene variant	33	ENST00000563303	?	?	7
LRRC36	upstream gene variant, downstream gene variant	2468	ENST00000569499	?	ENSP00000456814	4
LRRC36	upstream gene variant, downstream gene variant	72	ENST00000568804	?	ENSP00000464367	6
LRRC36	upstream gene variant, downstream gene variant	703	ENST00000567211	?	ENSP00000455211	2
PLEKHG4	upstream gene variant, downstream gene variant	1949	ENST00000393966	?	ENSP00000462601	3

PLEKHG4	upstream gene variant, downstream gene variant	1489	ENST00000562289	?	?	3
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000563969	?	ENSP00000457086	4
PLEKHG4	upstream gene variant	2561	ENST00000565899	?	ENSP00000455423	2
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000360461	NM_001129727.1, NM_015432.3	ENSP00000353646	4
PLEKHG4	upstream gene variant	3238	ENST00000565773	?	ENSP00000455876	2
PLEKHG4	upstream gene variant	3551	ENST00000562744	?	ENSP00000455622	2
PLEKHG4	upstream gene variant	3957	ENST00000568621	?	ENSP00000456873	1
PLEKHG4	upstream gene variant, downstream gene variant	1867	ENST00000567136	?	ENSP00000455228	3
PLEKHG4	upstream gene variant, downstream gene variant	1525	ENST00000427155	NM_001129728.1	ENSP00000401118	4
PLEKHG4	upstream gene variant, downstream gene variant	1817	ENST00000569875	?	ENSP00000464631	3
PLEKHG4	upstream gene variant	4000	ENST00000567938	?	ENSP00000455740	1
PLEKHG4	upstream gene variant, downstream gene variant	1531	ENST00000379344	NM_001129729.1	ENSP00000368649	3
PLEKHG4	upstream gene variant	2224	ENST00000562144	?	ENSP00000454348	2
PLEKHG4	upstream gene variant, downstream gene variant	1505	ENST00000450733	NM_001129731.1	ENSP00000398030	3
RN7SKP118	downstream gene variant, upstream gene variant	963	ENST00000364331	?	?	4
RNU1-123P	downstream gene variant, upstream gene variant	472	ENST00000458950	?	?	6
RP11-297D21.2	downstream gene variant, upstream gene variant	2887	ENST00000567261	?	?	4
RP11-297D21.4	upstream gene variant, downstream gene variant	1086	ENST00000602592	?	?	9
RP11-297D21.4	upstream gene variant	966	ENST00000602476	?	?	5
RP11-297D21.4	upstream gene variant, downstream gene variant	881	ENST00000602596	?	?	7
SLC9A5	downstream gene variant	2318	ENST00000564812	?	ENSP00000455058	2
SLC9A5	downstream gene variant	2186	ENST00000299798	NM_004594.2	ENSP00000299798	2
SLC9A5	downstream gene variant	2186	ENST00000563723	?	?	2
SLC9A5	downstream gene variant	2186	ENST00000564704	?	?	2
SLC9A5	downstream gene variant	3396	ENST00000566626	?	ENSP00000462404	2
TPPP3	downstream gene variant, upstream gene variant	2036	ENST00000564104	?	ENSP00000462435	6
TPPP3	downstream gene variant, upstream gene variant	471	ENST00000290942	NM_016140.3	ENSP00000290942	7
TPPP3	downstream gene variant, upstream gene variant	498	ENST00000562206	?	ENSP00000457275	7
TPPP3	downstream gene variant, upstream gene variant	454	ENST00000393957	NM_015964.3	ENSP00000377529	7
TPPP3	downstream gene variant, upstream gene variant	260	ENST00000561537	?	?	7
ZDHHC1	downstream gene variant, upstream gene variant	117	ENST00000565726	?	ENSP00000459264	6
ZDHHC1	downstream gene variant, upstream gene variant	15	ENST00000348579	NM_013304.2	ENSP00000340299	5
ZDHHC1	downstream gene variant, upstream gene variant	1203	ENST00000568650	?	?	4
ZDHHC1	downstream gene variant, upstream gene variant	1162	ENST00000567311	?	?	4
ZDHHC1	downstream gene variant, upstream gene variant	1083	ENST00000562122	?	?	7
ZDHHC1	downstream gene variant, upstream gene variant	21	ENST00000566075	?	?	5

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
AGRP	ENST00000290953	NM_001138.1	ENSP00000290953	E	gaA/gaG	1
HSD11B2	ENST00000326152	NM_000196.3	ENSP00000316786	2	2	2
TPPP3	ENST00000290942	NM_016140.3	ENSP00000290942	S	agT/agC	1
TPPP3	ENST00000393957	NM_015964.3	ENSP00000377529	S	agT/agC	1
TPPP3	ENST00000562206	?	ENSP00000457275	S	agT/agC	1
TPPP3	ENST00000564104	?	ENSP00000462435	S	agT/agC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V0D1	ENST00000565835	?	ENSP00000463328	16
ATP6V0D1	ENST00000426604	?	ENSP00000393910	8
ATP6V0D1	ENST00000564191	?	ENSP00000456695	16
ATP6V0D1	ENST00000564615	?	ENSP00000455802	6
ATP6V0D1	ENST00000290949	NM_004691.4	ENSP00000290949	16
ATP6V0D1	ENST00000567170	?	?	1
ATP6V0D1	ENST00000564788	?	ENSP00000455695	16
ATP6V0D1	ENST00000566322	?	ENSP00000455446	8
ATP6V0D1	ENST00000540149	?	ENSP00000441282	16
ATP6V0D1	ENST00000602876	?	ENSP00000473515	16
ATP6V0D1	ENST00000568101	?	ENSP00000457104	7
ATP6V0D1	ENST00000568298	?	?	10
ATP6V0D1	ENST00000563064	?	ENSP00000457105	16
ATP6V0D1	ENST00000561658	?	?	10
ATP6V0D1	ENST00000561852	?	ENSP00000457744	16
KCTD19	ENST00000570049	?	?	9
KCTD19	ENST00000566392	?	?	9
KCTD19	ENST00000562841	?	ENSP00000463249	2
KCTD19	ENST00000569333	?	?	9
KCTD19	ENST00000304372	NM_001100915.1	ENSP00000305702	8
KCTD19	ENST00000562860	?	?	4
KCTD19	ENST00000567976	?	ENSP00000458045	3
KCTD19	ENST00000568736	?	?	6
KCTD19	ENST00000562721	?	ENSP00000462122	6
KCTD19	ENST00000561625	?	?	6
KCTD19	ENST00000566295	?	ENSP00000462732	3
LRRC36	ENST00000435835	?	ENSP00000411122	18
LRRC36	ENST00000569228	?	?	28
LRRC36	ENST00000568804	?	ENSP00000464367	25
LRRC36	ENST00000567723	?	ENSP00000455799	19

LRRC36	ENST00000569552	?	ENSP00000454309	12
LRRC36	ENST00000567211	?	ENSP00000455211	13
LRRC36	ENST00000563189	NM_001161575.1	ENSP00000455103	17
LRRC36	ENST00000329956	NM_018296.5	ENSP00000329943	32
LRRC36	ENST00000561948	?	ENSP00000456347	25
LRRC36	ENST00000569499	?	ENSP00000456814	26
LRRC36	ENST00000565019	?	ENSP00000464675	33
LRRC36	ENST00000570075	?	ENSP00000454794	26
LRRC36	ENST00000568010	?	ENSP00000455018	18
LRRC36	ENST00000563303	?	?	16
LRRC36	ENST00000561821	?	ENSP00000456517	12
LRRC36	ENST00000566558	?	?	15
LRRC36	ENST00000567823	?	ENSP00000456164	19
PLEKHG4	ENST00000563969	?	ENSP00000457086	1
PLEKHG4	ENST00000379344	NM_001129729.1	ENSP00000368649	1
PLEKHG4	ENST00000393966	?	ENSP00000462601	1
PLEKHG4	ENST00000360461	NM_001129727.1, NM_015432.3	ENSP00000353646	1
PLEKHG4	ENST00000427155	NM_001129728.1	ENSP00000401118	1
PLEKHG4	ENST00000450733	NM_001129731.1	ENSP00000398030	1
RP11-297D21.4	ENST00000602476	?	?	3
RP11-297D21.4	ENST00000602596	?	?	2
TPPP3	ENST00000290942	NM_016140.3	ENSP00000290942	1
TPPP3	ENST00000393957	NM_015964.3	ENSP00000377529	1
TPPP3	ENST00000562206	?	ENSP00000457275	1
TPPP3	ENST00000564104	?	ENSP00000462435	1
ZDHHC1	ENST00000568650	?	?	9
ZDHHC1	ENST00000566075	?	?	1
ZDHHC1	ENST00000348579	NM_013304.2	ENSP00000340299	12
ZDHHC1	ENST00000567311	?	?	1
ZDHHC1	ENST00000562122	?	?	3
ZDHHC1	ENST00000565726	?	ENSP00000459264	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V0D1	ENST00000565835	?	ENSP00000463328	1
ATP6V0D1	ENST00000290949	NM_004691.4	ENSP00000290949	2
ATP6V0D1	ENST00000602876	?	ENSP00000473515	2
HSD11B2	ENST00000566606	?	ENSP00000473429	2
LRRC36	ENST00000567723	?	ENSP00000455799	3
LRRC36	ENST00000567211	?	ENSP00000455211	1
LRRC36	ENST00000565019	?	ENSP00000464675	2
LRRC36	ENST00000570075	?	ENSP00000454794	1

LRRC36	ENST00000568010	?	ENSP00000455018	2
LRRC36	ENST00000561821	?	ENSP00000456517	1
LRRC36	ENST00000567823	?	ENSP00000456164	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ATP6V0D1	ENST00000568620	?	2
ATP6V0D1	ENST00000563305	?	2
ATP6V0D1	ENST00000568298	?	2
HSD11B2	ENST00000567684	?	2
KCTD19	ENST00000570049	?	2
KCTD19	ENST00000566392	?	2
KCTD19	ENST00000569333	?	2
LRRC36	ENST00000569228	?	1
RNU1-123P	ENST00000458950	?	1
TPPP3	ENST00000561537	?	1