Locus 4 (rs9858542)

SNiPAcard

Block annotations

Block info

genomic range	chr3:49,385,417-49,731,861
block size	346,445 bp
variant count	107 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-3.849 – 1.994]	**gene(s) hit or close-by**	AC099668.1, AC099668.5, AMT, APEH, BSN, BSN-AS1, BSN-AS2, DAG1, GPX1, MST1, NICN1, RHOA, RHOA-IT1, RNA5SP130, RNF123, TCTA
phastCons	μ = 0.097 [0 – 1]	**eQTL gene(s)**	ACTBP13, AMT, APEH, C3orf62, CDHR4, CISH, DOCK3, FAM212A, GPX1, IP6K2, KLHDC8B, MIR5193, MST1, MST1R, NICN1, QRICH1, RBM6, RN7SL217P, RNF123, RP11-3B7.1, UBA7, USP4, WDR6
GERP++	μ = -0.427 [-10.1 – 4.85]	**potentially regulated gene(s)**	AMT, AMT, APEH, ARIH2, BSN, CAMKV, CDHR4, DAG1, DALRD3, IMPDH2, IP6K1, KLHDC8B, LAMB2, MON1A, MST1, NDUFAF3, QARS, QRICH1, RP11-3B7.1, SLC25A20, TRAIP, UBA7, USP4
CADD score	μ = 4.712 [0.004 – 22.1]	**disease gene(s)**	AMT, KLHDC8B, LAMB2, NDUFAF3, SLC25A20, QARS, DAG1, CISH, MST1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Educational attainment	$<5.00\times10^{-9}$	GWAS Catalog	25201988	1
Ulcerative colitis	$<8.00\times10^{-7}$	GWAS Catalog	25082827	1
Inflammatory bowel disease	$<1.00\times10^{-47}$	GWAS Catalog	23128233	1
Ulcerative colitis	$<2.00\times10^{-17}$	GWAS Catalog	21297633	1
Primary sclerosing cholangitis	$<1.00\times10^{-16}$	GWAS Catalog	21151127	1
Crohn's disease (time to surgery)	$<6.00\times10^{-17}$	GWAS Catalog	21102463	1
Ulcerative colitis	$<4.00\times10^{-9}$	GWAS Catalog	20228799	1
Ulcerative colitis	$<7.00\times10^{-9}$	GWAS Catalog	19915572	1
Crohn's disease (time to surgery)	$<1.00\times10^{-12}$	GWAS Catalog	18587394	1
Crohn's disease (time to surgery)	$<4.00\times10^{-8}$	GWAS Catalog	17554300	1
Crohn's disease (time to surgery)	$<5.00\times10^{-8}$	GWAS Catalog	17554261	1
benzoate	6.67×10^{-5}	Metabolomics GWAS Server	24816252	2

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Clinvar: phenotype not specified	likely benign	ClinVar	RCV000116863.2	1
Clinvar: phenotype not specified	not provided	ClinVar	RCV000024450.1	1
Clinvar: phenotype not specified	not provided	ClinVar	RCV000024446.1	1
Glutathione peroxidase polymorphism	benign	ClinVar	RCV000017462.1	1
?	HGMD curated	HGMD	CR045943	1
?	HGMD curated	HGMD	CR045942	1
?	HGMD curated	HGMD	CM087622	1

?	HGMD curated	HGMD	CM061794	HGMD	1
GLUTATHIONE PEROXIDASE POLYMORPHISM	OMIM curated	OMIM	MIM:138320	OMIM	1

Disease gene annotation

gene	trait	source DB	source entry/link	
AMT	GLYCINE ENCEPHALOPATHY	OMIM	MIM:605899	OMIM
KLHDC8B	LYMPHOMA, HODGKIN, CLASSIC	OMIM	MIM:236000	OMIM
LAMB2	NEPHROTIC SYNDROME, TYPE 5, WITH OR WITHOUT OCULAR [...]	OMIM	MIM:614199	OMIM
LAMB2	PIERSON SYNDROME	OMIM	MIM:609049	OMIM
NDUFAF3	MITOCHONDRIAL COMPLEX I DEFICIENCY	OMIM	MIM:252010	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OMIM	MIM:212138	OMIM
QARS	MICROCEPHALY, PROGRESSIVE, WITH SEIZURES AND CEREBRAL AND [...]	OMIM	MIM:615760	OMIM
DAG1	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE C, [...]	OMIM	MIM:613818	OMIM
CISH	MYCOBACTERIUM TUBERCULOSIS, SUSCEPTIBILITY TOMYCOBACTERIUM TUBERCULOSIS, [...]	OMIM	MIM:607948	OMIM
CISH	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
CISH	BACTEREMIA, SUSCEPTIBILITY TO, 2	OMIM	MIM:614383	OMIM
AMT	GLYCINE ENCEPHALOPATHY	DECIPHER	MIM:605899	OMIM
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	DECIPHER	MIM:212138	OMIM
QARS	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy	DECIPHER	MIM:615760	OMIM
DAG1	muscular dystrophy-dystroglycanopathy limb-girdle type C7 (MDDGC7)	DECIPHER	MIM:613818	OMIM
AMT	Infantile glycine encephalopathy	OrphaNet	OrphaNet:289860	orphanet
AMT	Neonatal glycine encephalopathy	OrphaNet	OrphaNet:289857	orphanet
AMT	Atypical glycine encephalopathy	OrphaNet	OrphaNet:289863	orphanet
MST1	Primary sclerosing cholangitis	OrphaNet	OrphaNet:171	orphanet
LAMB2	Synaptic congenital myasthenic syndromes	OrphaNet	OrphaNet:98915	orphanet
LAMB2	LAMB-2-related infantile-onset nephrotic syndrome	OrphaNet	OrphaNet:306507	orphanet
LAMB2	PIERSON SYNDROME	OrphaNet	OrphaNet:2670	orphanet
NDUFAF3	Isolated NADH-CoQ reductase deficiency	OrphaNet	OrphaNet:2609	orphanet
SLC25A20	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	OrphaNet	OrphaNet:159	orphanet
DAG1	Autosomal recessive limb-girdle muscular dystrophy type 2P	OrphaNet	OrphaNet:280333	orphanet
DAG1	Muscle-eye-brain disease with bilateral multicystic leucodystrophy	OrphaNet	OrphaNet:370997	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)
BSN	missense variant	ENST00000296452	NM_003458.3	ENSP00000296452	T/A	Acc/Gcc	?	?	1
GPX1	missense variant	ENST00000620890	?	ENSP00000478837	L/P	cTc/cCc	?	?	1
GPX1	missense variant	ENST00000419783	NM_000581.2	ENSP00000407375	L/P	cTc/cCc	?	?	1
MST1	missense variant	ENST00000449682	NM_020998.3	ENSP00000414287	C/R	Tgc/Cgc	?	?	1
MST1	missense variant	ENST00000448220	?	ENSP00000394756	C/R	Tgc/Cgc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GPX1	?	ENSG00000233276	muscularis mucosae	1.54×10^{-8} (p-value)	GTEx Portal V6	106

RBM6	?	ENSG00000004534	muscularis mucosae	2.15×10-6 (p-value)	GTEx Portal V6	102
NICN1	?	ENSG00000145029	muscularis mucosae	4.84×10-7 (p-value)	GTEx Portal V6	106
RNF123	?	ENSG00000164068	muscularis mucosae	7.42×10-6 (p-value)	GTEx Portal V6	48
IP6K2	?	ENSG00000068745	muscularis mucosae	3.61×10-5 (p-value)	GTEx Portal V6	1
AMT	?	ENSG00000145020	muscularis mucosae	2.79×10-5 (p-value)	GTEx Portal V6	3
RBM6	?	ENSG00000004534	lung	1.43×10-7 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	lung	2.85×10-7 (p-value)	GTEx Portal V6	105
AMT	?	ENSG00000145020	lung	4.83×10-7 (p-value)	GTEx Portal V6	104
AMT	?	ENSG00000145020	transformed fibroblasts	6.52×10-7 (p-value)	GTEx Portal V6	105
IP6K2	?	ENSG00000068745	transformed fibroblasts	2.39×10-8 (p-value)	GTEx Portal V6	80
APEH	?	ENSG00000164062	transformed fibroblasts	6.85×10-13 (p-value)	GTEx Portal V6	106
RNF123	?	ENSG00000164068	transformed fibroblasts	1.48×10-5 (p-value)	GTEx Portal V6	39
RBM6	?	ENSG00000004534	transformed fibroblasts	6.37×10-6 (p-value)	GTEx Portal V6	11
GPX1	?	ENSG00000233276	tibial artery	8.26×10-8 (p-value)	GTEx Portal V6	105
RBM6	?	ENSG00000004534	tibial artery	6.49×10-8 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	tibial artery	1.09×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	tibial artery	2.71×10-10 (p-value)	GTEx Portal V6	106
IP6K2	?	ENSG00000068745	tibial artery	3.53×10-5 (p-value)	GTEx Portal V6	1
RNF123	?	ENSG00000164068	tibial artery	8.74×10-6 (p-value)	GTEx Portal V6	96
RBM6	?	ENSG00000004534	breast	9.52×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	breast	1.22×10-5 (p-value)	GTEx Portal V6	2
RBM6	?	ENSG00000004534	blood	4.69×10-8 (p-value)	GTEx Portal V6	106
UBA7	?	ENSG00000182179	blood	3.32×10-8 (p-value)	GTEx Portal V6	104
GPX1	?	ENSG00000233276	thyroid	2.62×10-8 (p-value)	GTEx Portal V6	105
RBM6	?	ENSG00000004534	thyroid	1.97×10-10 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	thyroid	3.22×10-9 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	thyroid	1.04×10-6 (p-value)	GTEx Portal V6	89
RNF123	?	ENSG00000164068	thyroid	6.41×10-6 (p-value)	GTEx Portal V6	87
MST1	?	ENSG00000173531	thyroid	4.85×10-8 (p-value)	GTEx Portal V6	101
FAM212A	?	ENSG00000185614	skeletal muscle	7.82×10-15 (p-value)	GTEx Portal V6	106
IP6K2	?	ENSG00000068745	skeletal muscle	2.44×10-5 (p-value)	GTEx Portal V6	3
RBM6	?	ENSG00000004534	skeletal muscle	2.87×10-5 (p-value)	GTEx Portal V6	2
UBA7	?	ENSG00000182179	transverse colon	9.12×10-9 (p-value)	GTEx Portal V6	106
RBM6	?	ENSG00000004534	transverse colon	9.71×10-6 (p-value)	GTEx Portal V6	2
UBA7	?	ENSG00000182179	unexposed skin	1.69×10-5 (p-value)	GTEx Portal V6	50
MST1	?	ENSG00000173531	unexposed skin	3.70×10-7 (p-value)	GTEx Portal V6	82
RBM6	?	ENSG00000004534	unexposed skin	3.48×10-6 (p-value)	GTEx Portal V6	7

RBM6	?	ENSG00000004534	sun exposed skin	1.84×10-10 (p-value)	GTEx Portal V6	106
KLHDC8B	?	ENSG00000185909	sun exposed skin	1.22×10-5 (p-value)	GTEx Portal V6	32
ACTBP13	?	ENSG00000234667	sun exposed skin	2.94×10-6 (p-value)	GTEx Portal V6	43
APEH	?	ENSG00000164062	sun exposed skin	2.71×10-5 (p-value)	GTEx Portal V6	1
MST1	?	ENSG00000173531	sun exposed skin	2.34×10-6 (p-value)	GTEx Portal V6	32
WDR6	?	ENSG00000178252	sun exposed skin	3.57×10-5 (p-value)	GTEx Portal V6	1
DOCK3	?	ENSG00000088538	sun exposed skin	3.30×10-5 (p-value)	GTEx Portal V6	2
GPX1	?	ENSG00000233276	aorta	5.78×10-6 (p-value)	GTEx Portal V6	33
NICN1	?	ENSG00000145029	aorta	3.82×10-7 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	aorta	3.13×10-6 (p-value)	GTEx Portal V6	87
RBM6	?	ENSG00000004534	aorta	4.07×10-5 (p-value)	GTEx Portal V6	1
RBM6	?	ENSG00000004534	subcutaneous adipocytes	2.38×10-9 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	subcutaneous adipocytes	3.02×10-10 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	subcutaneous adipocytes	4.25×10-11 (p-value)	GTEx Portal V6	106
APEH	?	ENSG00000164062	subcutaneous adipocytes	7.03×10-7 (p-value)	GTEx Portal V6	105
RNF123	?	ENSG00000164068	subcutaneous adipocytes	3.52×10-6 (p-value)	GTEx Portal V6	98
MST1R	?	ENSG00000164078	subcutaneous adipocytes	2.36×10-5 (p-value)	GTEx Portal V6	1
RBM6	?	ENSG00000004534	visceral adipocytes	5.54×10-7 (p-value)	GTEx Portal V6	102
MIR5193	?	ENSG00000263506	tibial nerve	4.13×10-7 (p-value)	GTEx Portal V6	97
RBM6	?	ENSG00000004534	tibial nerve	1.57×10-8 (p-value)	GTEx Portal V6	106
NICN1	?	ENSG00000145029	tibial nerve	7.68×10-8 (p-value)	GTEx Portal V6	105
AMT	?	ENSG00000145020	tibial nerve	7.07×10-7 (p-value)	GTEx Portal V6	104
MST1	?	ENSG00000173531	tibial nerve	2.47×10-8 (p-value)	GTEx Portal V6	103
RNF123	?	ENSG00000164068	tibial nerve	1.40×10-6 (p-value)	GTEx Portal V6	101
WDR6	?	ENSG00000178252	esophagus mucosa	7.08×10-7 (p-value)	GTEx Portal V6	24
RBM6	?	ENSG00000004534	esophagus mucosa	6.37×10-9 (p-value)	GTEx Portal V6	106
AMT	?	ENSG00000145020	esophagus mucosa	1.28×10-6 (p-value)	GTEx Portal V6	72
FAM212A	?	ENSG00000185614	esophagus mucosa	1.66×10-9 (p-value)	GTEx Portal V6	106
APEH	?	ENSG00000164062	esophagus mucosa	1.64×10-7 (p-value)	GTEx Portal V6	103
UBA7	?	ENSG00000182179	esophagus mucosa	2.67×10-5 (p-value)	GTEx Portal V6	1
QRICH1	?	ENSG00000198218	pituitary	1.34×10-6 (p-value)	GTEx Portal V6	67
GPX1	?	ENSG00000233276	cerebellar hemisphere	4.45×10-7 (p-value)	GTEx Portal V6	105
GPX1	?	ENSG00000233276	cerebellum	1.29×10-7 (p-value)	GTEx Portal V6	105
MST1R	?	ENSG00000164078	cerebellum	4.34×10-7 (p-value)	GTEx Portal V6	40
RNF123	?	ENSG00000164068	cerebellum	1.83×10-5 (p-value)	GTEx Portal V6	2
GPX1	?	ENSG00000233276	frontal cortex	6.00×10-6 (p-value)	GTEx Portal V6	62
RNF123	?	ENSG00000164068	atrial appendage	2.55×10-7 (p-value)	GTEx Portal V6	97

RBM6	?	ENSG00000004534	atrial appendage	2.51×10-5 (p-value)	GTEx Portal V6	1
GPX1	?	ENSG00000233276	atrial appendage	1.44×10-5 (p-value)	GTEx Portal V6	2
RBM6	ENST00000442092	ILMN_1738239	monocyte	1.28×10-6 (p-value)	Fairfax et al.	15
RBM6	ENST00000266022		b-cell	2.89×10-6 (p-value)	Fairfax et al.	15
RBM6	ENST00000454079					
RBM6	ENST00000422955					
RBM6	ENST00000419610					
RBM6	ENST00000443081					
RBM6	ENST00000421682					
USP4	ENST00000351842	ILMN_1798712	monocyte	7.21×10-8 (p-value)	Fairfax et al.	14
USP4	ENST00000483212					
C3orf62	ENST00000343010					
USP4	ENST00000485450					
UBA7	ENST00000497908	ILMN_1794612	b-cell	8.34×10-11 (p-value)	Fairfax et al.	1
UBA7	ENST00000333486					
MIR5193	ENST00000584510					
CDHR4	?	ENSG00000187492	testis	1.64×10-5 (p-value)	GTEx Portal V6	64
IP6K2	?	ENSG00000068745	testis	4.07×10-5 (p-value)	GTEx Portal V6	1
RN7SL217P	?	ENSG00000264706	testis	2.24×10-5 (p-value)	GTEx Portal V6	35
WDR6	ENST00000615452	ILMN_1669484	skin	9.77×10-15 (p-value)	MuTHER consortium	38
WDR6	ENST00000452875					
WDR6	ENST00000471162					
WDR6	ENST00000610967					
WDR6	ENST00000395474					
WDR6	ENST00000492780					
WDR6	ENST00000608424					
WDR6	ENST00000420783					
APEH	ENST00000480772	ILMN_1718023	blood	2.51×10-5 (p-value)	MuTHER consortium	35
APEH	ENST00000469362					
APEH	ENST00000296456					
APEH	ENST00000438011					
USP4	ENST00000351842	ILMN_1798712	adipocyte	7.71×10-6 (p-value)	MuTHER consortium	31
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
RP11-3B7.1	ENST00000622888	ILMN_1807244	adipocyte	1.14×10-5 (p-value)	MuTHER consortium	17
RP11-3B7.1	ENST00000440528					
KLHDC8B	ENST00000332780	ILMN_1695246	blood	2.66×10-5 (p-value)	Westra et al.	10
KLHDC8B	ENST00000462582					
FAM212A	ENST00000333323	ILMN_1690454	blood	3.10×10-6 (p-value)	Westra et al.	2
USP4	ENST00000351842	ILMN_1798712	monocyte	1.65×10-13 (p-value)	Zeller et al.	7
USP4	ENST00000483212					
USP4	ENST00000485450					
C3orf62	ENST00000343010					
CISH	?	ENSG00000114737	blood	2.90×10-2 (q-value)	SeeQTL DB (HapMap)	9
MST1	?	ENSG00000173531	brain	4.01×10-2 (q-value)	SeeQTL DB (Myers et al.)	1
RBM6	?	ENSG00000004534	sigmoid colon	1.88×10-5 (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)
ENCE00000338841	1	ENCP00000036479	LAMB2
		ENCP00000036455	DALRD3 NDUFAF3
		ENCP00000036491	RP11-3B7.1
		ENCP00000036487	KLHDC8B
		ENCP00000036545	UBA7
		ENCP00000036459	IMPDH2
		ENCP00000036454	NDUFAF3
		ENCP00000036522	APEH
		ENCP00000036544	UBA7
		ENCP00000036463	QRICH1
		ENCP00000036525	MST1
		ENCP00000036498	USP4
		ENCP00000036434	ARIH2
		ENCP00000036429	SLC25A20
		ENCP00000036496	USP4
		ENCP00000036461	IMPDH2
		ENCP00000036458	DALRD3
		ENCP00000036546	UBA7
ENCE00000338858	1	ENCP00000036479	LAMB2
		ENCP00000036455	DALRD3 NDUFAF3
		ENCP00000036487	KLHDC8B
		ENCP00000036459	IMPDH2
		ENCP00000036454	NDUFAF3
		ENCP00000036522	APEH
		ENCP00000036544	UBA7
		ENCP00000036463	QRICH1
		ENCP00000036525	MST1
		ENCP00000036534	IP6K1
		ENCP00000036498	USP4
		ENCP00000036434	ARIH2
		ENCP00000036496	USP4
		ENCP00000036551	CAMKV
		ENCP00000036461	IMPDH2
		ENCP00000036472	QARS
		ENCP00000036458	DALRD3
		ENCP00000036546	UBA7
ENCE00000338949	1	ENCP00000036468	QARS
		ENCP00000036469	QARS
ENCE00000339059	1	ENCP00000036562	MON1A
		ENCP00000036549	TRAIP
		ENCP00000036518	BSN
		ENCP00000036509	AMT AMT
		ENCP00000036540	CDHR4
		ENCP00000036552	CAMKV
		ENCP00000036515	DAG1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363628 (promoter flanking region)	4	NHLF	DNase1, H3K27ac
		HSMMtube	DNase1
		blood (K562)	Gata2, H3K27ac, DNase1
		skin (NHDF-AD)	H3K4me1, DNase1, H3K4me2
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	PolII, H3K4me2, H3K27ac, H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3, H3K9ac
		endothelium (HUVEC)	Cjun, DNase1, H3K27ac, PolII
		liver (HepG2)	H3K4me1, H3K4me2, FOXA1
		blood (GM12878)	PU1, PolII, MEF2A, H2AZ, H3K4me1, Tcf12, DNase1, Yy1, H3K4me3, Pax5, H3K27ac, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K4me3
		skin (NHEK)	H3K4me1
ENSR00001363631 (promoter)	3	embryonic stem cell (H1ESC)	DNase1, Rad21, TAF7, H3K27ac, ATF3, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3, H3K27me3, PolII
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, H3K36me3
		muscle (HSMM)	H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K36me3
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, USF1, Cmyc, PolII, DNase1, ELF1, H3K79me2, H3K4me3
		lung (IMR90)	H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, DNase1, H3K36me3, H3K4me3
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, Cmyc, H3K79me2, Gabp, PolII, H3K36me3, H3K4me3, Max, TAF1, H3K9ac, Jund, H3K4me2, H3K27ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, H3K9ac, H3K4me1, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001674821 (enhancer)	1	HSMMtube	H3K79me2
		Osteobl	H3K27ac
		blood (K562)	H3K79me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K79me2
		A549	H3K36me3
ENSR00001674822 (enhancer)	1	HSMMtube	H3K79me2
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2, H3K36me3
		cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		nervous (NH-A)	H4K20me1
		A549	H3K36me3
ENSR00001363642 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, TAF7, Rad21, H3K36me3, PolII
		HSMMtube	H3K79me2, H3K9ac, H4K20me1, H3K4me2, H3K27ac, H3K4me3, DNase1
		blood (K562)	H3K27ac, Max, H3K9ac, H3K79me2, HEY1, H2AZ, TAF1, PolII, H3K4me2, DNase1, H3K4me3

		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1, H3K4me2, H3K4me1, H4K20me1, PolII, H3K79me2, TAF1
		blood (GM12878)	PolII, MEF2A, H2AZ, DNase1, Yy1, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		lung (IMR90)	H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H2AK5ac, H3K9ac, H3K79me2, H3K14ac, DNase1, H4K8ac, H4K20me1
		nervous (NH-A)	H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, H3K4me1, H3K4me2
		breast (HMEC)	H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K36me3, H3K4me3, DNase1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, PolII, DNase1, Cmyc, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001363643 (promoter flanking region)	2	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, DNase1
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K36me3
		breast (HMEC)	CTCF
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	PolII, Rad21, CTCF, H3K36me3
		blood (GM12878)	EBF1, PolII, DNase1, CTCF, Rad21, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
ENSR00001363645 (enhancer)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	Jund, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	FOSL2, Jund, PolII, H3K4me1
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF
ENSR00001363654 (promoter flanking region)	2	cervix (HeLa-S3)	H3K27ac, H3K4me1, DNase1, CTCF, Jund
		embryonic stem cell (H1ESC)	Rad21, DNase1
		liver (HepG2)	H3K4me1
		HSMMtube	DNase1
		A549	CTCF
		muscle (HSMM)	DNase1
ENSR00001479030 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	EBF1, BATF, MEF2A
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001363667 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	DNase1, H3K4me1, H3K4me2, H3K27ac
		breast (HMEC)	H3K27ac
ENSR00001479032 (promoter flanking region)	5	embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3, H3K27ac, H3K9ac, DNase1

		blood (K562)	DNase1, H3K27ac, H3K9ac, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3, H3K27ac, H3K4me1, H3K4me2, HNF4A, H3K9ac
		lung (IMR90)	DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me2, H3K27ac
		NHLF	DNase1, H3K27ac
		Osteobl	H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
		cervix (HeLa-S3)	H3K4me1, H3K36me3, DNase1, H3K27ac, H3K4me2, PolII, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac
		endothelium (HUVEC)	DNase1
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac, H3K36me3
ENSR00001479035 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27ac, DNase1
		blood (DND-41)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	H3K27ac
ENSR00001674842 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me1
		HSMMtube	H3K27ac
ENSR00001363673 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K27me3, Rad21, Nrsf, Sin3Ak20, Yy1, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H2AZ, H3K27me3, DNase1, H3K4me2, H3K4me3
		blood (K562)	H3K4me3, DNase1, Sin3Ak20, Max, H3K9ac, H2AZ, Nrsf, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2
		liver (HepG2)	H3K27me3, DNase1, H3K4me3, H3K9ac, Nrsf, PolII, Sin3Ak20, TAF1, H3K4me1, H2AZ, H3K4me2
		lung (IMR90)	H3K4me3, DNase1, H3K4me2, H3K27me3
		blood (GM12878)	DNase1, Nrsf, H2AZ, Yy1, H3K4me3, H3K4me2, H3K27me3
		nervous (NH-A)	H3K4me3, H3K27me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K27me3, H3K4me3, H3K4me2
		NHLF	H3K27me3, H3K9ac, H3K4me3
		Osteobl	H3K27me3, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	Ini1, H3K9ac, Nrsf, H3K4me2, H3K27me3, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	PolII, H3K27me3, DNase1
		A549	H3K4me3, H3K4me2, H3K9ac, H3K27me3
ENSR00001479043 (enhancer)	2	lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHEK)	H3K4me1, DNase1
ENSR00001363687 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	DNase1, Max, MEF2A, PolII
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		cervix (HeLa-S3)	DNase1, Cjun, Jund, H3K4me1
		endothelium (HUVEC)	H3K36me3, H3K27me3
		liver (HepG2)	DNase1
		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me1, DNase1
ENSR00001363693 (promoter flanking region)	1	cervix (HeLa-S3)	CTCF
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	DNase1, H3K4me1, FOXA1, CTCF
		lung (IMR90)	H3K27me3

		Osteobl	H3K27me3
		blood (K562)	CTCF
ENSR00001479046 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K27me3, PolII, Gabp, Sin3Ak20, H3K9ac, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K4me3
		HSMMtube	H3K4me3, H2AZ, DNase1, H3K4me2, H3K27ac
		blood (K562)	H3K79me2, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, Ini1, Egr1, H3K27ac, Max, CTCF, H3K9ac
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, H3K4me2, H3K27ac, H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, H3K4me3, H3K36me3, Cmyc, H3K27ac, Gabp, CTCF, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	H3K4me2, H3K36me3, H3K4me3, H3K27ac, DNase1
		blood (GM12878)	EBF1, PolII, H2AZ, DNase1, CTCF, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H3K4me3, H3K36me3, CTCF, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, CTCF, H3K27ac, Max, H3K4me3, Gabp, PolII, Cmyc, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, Max, H3K4me3, CTCF, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, CTCF, DNase1, H3K27ac, H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC099668.1	upstream gene variant, downstream gene variant	211	ENST00000630332	?	?	3
AC099668.5	downstream gene variant	115	ENST00000563780	?	?	3
AMT	downstream gene variant, upstream gene variant	99	ENST00000465925	?	?	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000273588	NM_000481.3	ENSP00000273588	4
AMT	downstream gene variant, upstream gene variant	1002	ENST00000473163	?	?	4
AMT	downstream gene variant, upstream gene variant	851	ENST00000399379	?	ENSP00000399943	4
AMT	downstream gene variant, upstream gene variant	305	ENST00000491800	?	?	4
AMT	downstream gene variant, upstream gene variant	767	ENST00000478594	?	?	5
AMT	downstream gene variant, upstream gene variant	1657	ENST00000462048	?	?	5
AMT	downstream gene variant, upstream gene variant	100	ENST00000538581	NM_001164711.1	ENSP00000443200	4
AMT	downstream gene variant, upstream gene variant	721	ENST00000476127	?	?	4
AMT	downstream gene variant, upstream gene variant	99	ENST00000427987	?	ENSP00000403821	4
AMT	downstream gene variant, upstream gene variant	851	ENST00000495436	?	?	4
AMT	downstream gene variant, upstream gene variant	1216	ENST00000430521	?	ENSP00000388068	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000458307	NM_001164710.1	ENSP00000415619	4
AMT	downstream gene variant, upstream gene variant	99	ENST00000395338	NM_001164712.1	ENSP00000378747	4
AMT	downstream gene variant, upstream gene variant	100	ENST00000476226	?	?	4
AMT	downstream gene variant, upstream gene variant	1531	ENST00000485108	?	?	5
AMT	downstream gene variant, upstream gene variant	1449	ENST00000493046	?	?	5
AMT	downstream gene variant, upstream gene variant	735	ENST00000461210	?	?	5
AMT	downstream gene variant, upstream gene variant	819	ENST00000498571	?	?	4
AMT	downstream gene variant, upstream gene variant	725	ENST00000487589	?	?	5

AMT	downstream gene variant, upstream gene variant	1287	ENST00000480957	?	?	5
AMT	downstream gene variant, upstream gene variant	1641	ENST00000476828	?	?	5
APEH	upstream gene variant	968	ENST00000449966	?	ENSP00000414369	1
APEH	upstream gene variant, downstream gene variant	1314	ENST00000446089	?	ENSP00000410149	4
APEH	upstream gene variant, downstream gene variant	440	ENST00000483715	?	?	2
APEH	upstream gene variant, downstream gene variant	286	ENST00000462439	?	?	2
APEH	upstream gene variant, downstream gene variant	1154	ENST00000469362	?	ENSP00000438180	4
APEH	upstream gene variant, downstream gene variant	1339	ENST00000447436	?	ENSP00000401871	4
APEH	upstream gene variant, downstream gene variant	276	ENST00000491799	?	?	2
APEH	upstream gene variant	1333	ENST00000463995	?	?	1
APEH	upstream gene variant, downstream gene variant	830	ENST00000447541	?	ENSP00000404353	3
APEH	upstream gene variant, downstream gene variant	757	ENST00000438011	?	ENSP00000415862	3
APEH	upstream gene variant, downstream gene variant	1806	ENST00000482301	?	?	5
APEH	upstream gene variant, downstream gene variant	1321	ENST00000442186	?	ENSP00000402365	2
APEH	upstream gene variant	1440	ENST00000457042	?	ENSP00000410366	1
APEH	upstream gene variant, downstream gene variant	136	ENST00000296456	NM_001640.3	ENSP00000296456	3
APEH	upstream gene variant, downstream gene variant	560	ENST00000463616	?	?	2
APEH	upstream gene variant, downstream gene variant	305	ENST00000480772	?	?	3
BSN	upstream gene variant, downstream gene variant	1152	ENST00000296452	NM_003458.3	ENSP00000296452	3
BSN	upstream gene variant, downstream gene variant	428	ENST00000467456	?	?	4
BSN-AS1	downstream gene variant, upstream gene variant	2582	ENST00000442384	?	?	4
DAG1	upstream gene variant	3617	ENST00000452060	?	ENSP00000410145	1
DAG1	upstream gene variant, downstream gene variant	1856	ENST00000479935	?	?	4
DAG1	upstream gene variant	3627	ENST00000466701	?	?	1
DAG1	upstream gene variant	2051	ENST00000469139	?	?	1
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000545947	NM_001177634.2	ENSP00000442600	3
DAG1	upstream gene variant	3692	ENST00000496474	?	?	1
DAG1	upstream gene variant, downstream gene variant	1556	ENST00000461492	?	?	3
DAG1	upstream gene variant, downstream gene variant	1760	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	3
DAG1	upstream gene variant	3657	ENST00000430636	?	ENSP00000401805	1
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000539901	NM_001177644.2	ENSP00000439334	3
DAG1	upstream gene variant, downstream gene variant	1936	ENST00000421560	?	ENSP00000412067	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000515359	NM_001177643.2	ENSP00000440705	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	3
DAG1	upstream gene variant	3615	ENST00000452317	?	ENSP00000387859	1
DAG1	upstream gene variant	3615	ENST00000435508	?	ENSP00000415321	1
DAG1	upstream gene variant, downstream gene variant	248	ENST00000461987	?	?	4

DAG1	upstream gene variant	3654	ENST00000419218	?	ENSP00000402443	1
DAG1	upstream gene variant, downstream gene variant	1903	ENST00000431960	?	ENSP00000388833	3
DAG1	upstream gene variant, downstream gene variant	1757	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	3
DAG1	upstream gene variant	3638	ENST00000428779	?	ENSP00000401382	1
DAG1	upstream gene variant	667	ENST00000475424	?	?	2
DAG1	upstream gene variant	3474	ENST00000418588	?	ENSP00000405859	1
GPX1	downstream gene variant, upstream gene variant	207	ENST00000496791	?	?	5
GPX1	downstream gene variant, upstream gene variant	327	ENST00000419783	NM_000581.2	ENSP00000407375	5
GPX1	downstream gene variant, upstream gene variant	41	ENST00000419349	NM_201397.1	ENSP00000391316	6
GPX1	downstream gene variant, upstream gene variant	569	ENST00000620890	?	ENSP00000478837	5
MST1	downstream gene variant	2675	ENST00000481055	?	?	3
MST1	downstream gene variant	496	ENST00000497359	?	?	3
MST1	downstream gene variant, upstream gene variant	2377	ENST00000494828	?	?	4
MST1	downstream gene variant, upstream gene variant	1803	ENST00000492370	?	?	4
MST1	downstream gene variant	226	ENST00000484144	?	?	2
MST1	downstream gene variant, upstream gene variant	3448	ENST00000491943	?	?	4
MST1	downstream gene variant	1651	ENST00000448220	?	ENSP00000394756	1
MST1	downstream gene variant	1042	ENST00000480268	?	?	3
MST1	downstream gene variant	1680	ENST00000492329	?	?	1
MST1	downstream gene variant	1753	ENST00000493836	?	?	1
MST1	downstream gene variant	1380	ENST00000489007	?	?	3
MST1	downstream gene variant	373	ENST00000484269	?	?	3
MST1	downstream gene variant, upstream gene variant	1520	ENST00000484673	?	?	4
MST1	downstream gene variant	1318	ENST00000468847	?	?	3
MST1	downstream gene variant, upstream gene variant	2603	ENST00000490966	?	?	4
MST1	downstream gene variant, upstream gene variant	1680	ENST00000488350	?	?	3
MST1	downstream gene variant	373	ENST00000481930	?	?	3
MST1	downstream gene variant, upstream gene variant	1651	ENST00000449682	NM_020998.3	ENSP00000414287	2
MST1	downstream gene variant	479	ENST00000498021	?	?	3
MST1	downstream gene variant	1680	ENST00000479115	?	?	1
MST1	downstream gene variant	1117	ENST00000494809	?	?	3
NICN1	downstream gene variant, upstream gene variant	3941	ENST00000494057	?	?	2
NICN1	downstream gene variant	4376	ENST00000461015	?	?	1
NICN1	downstream gene variant, upstream gene variant	3947	ENST00000422593	?	?	2
NICN1	downstream gene variant, upstream gene variant	2071	ENST00000423832	?	ENSP00000413115	3
NICN1	downstream gene variant, upstream gene variant	2025	ENST00000615713	?	ENSP00000478287	3
NICN1	downstream gene variant, upstream gene variant	2024	ENST00000273598	NM_032316.3	ENSP00000273598	3

NICN1	downstream gene variant, upstream gene variant	3255	ENST00000436744	?	ENSP00000402335	2
NICN1	downstream gene variant, upstream gene variant	3153	ENST00000497742	?	?	2
RHOA	downstream gene variant, upstream gene variant	691	ENST00000445425	?	ENSP00000408402	6
RHOA	downstream gene variant, upstream gene variant	965	ENST00000454011	?	ENSP00000394483	6
RHOA	downstream gene variant, upstream gene variant	218	ENST00000418115	NM_001664.2	ENSP00000400175	6
RHOA	upstream gene variant	18	ENST00000265538	?	?	4
RHOA	downstream gene variant, upstream gene variant	919	ENST00000431929	?	?	6
RHOA	downstream gene variant, upstream gene variant	1046	ENST00000422781	?	ENSP00000413587	6
RHOA-IT1	downstream gene variant, upstream gene variant	725	ENST00000428083	?	?	5
RNA5SP130	downstream gene variant	453	ENST00000364431	?	?	1
TCTA	downstream gene variant	2975	ENST00000488385	?	?	3
TCTA	downstream gene variant	959	ENST00000493381	?	?	4
TCTA	downstream gene variant	3268	ENST00000497786	?	?	3
TCTA	downstream gene variant	204	ENST00000273590	NM_022171.2	ENSP00000273590	4
TCTA	downstream gene variant	3129	ENST00000482193	?	?	3
TCTA	downstream gene variant	1136	ENST00000487432	?	?	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
AMT	ENST00000430521	?	ENSP00000388068	R	cgA/cgG	1
AMT	ENST00000427987	?	ENSP00000403821	R	cgA/cgG	1
AMT	ENST00000538581	NM_001164711.1	ENSP00000443200	R	cgA/cgG	1
AMT	ENST00000458307	NM_001164710.1	ENSP00000415619	R	cgA/cgG	1
AMT	ENST00000273588	NM_000481.3	ENSP00000273588	R	cgA/cgG	1
AMT	ENST00000395338	NM_001164712.1	ENSP00000378747	R	cgA/cgG	1
APEH	ENST00000296456	NM_001640.3	ENSP00000296456	Y	taC/taT	1
APEH	ENST00000449966	?	ENSP00000414369	Y	taC/taT	1
APEH	ENST00000457042	?	ENSP00000410366	Y	taC/taT	1
APEH	ENST00000438011	?	ENSP00000415862	Y	taC/taT	1
APEH	ENST00000442186	?	ENSP00000402365	Y	taC/taT	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	2	2	2
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	H	caT/caC	1
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	H	caT/caC	1
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	H	caT/caC	1
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	H	caT/caC	1
DAG1		NM_001177644.2	ENSP00000439334	H	caT/caC	1

	ENST00000539901					
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	H	caT/caC	1
MST1	ENST00000449682	NM_020998.3	ENSP00000414287	R	cgT/cgG	1
MST1	ENST00000448220	?	ENSP00000394756	R	cgT/cgG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AMT	ENST00000399379	?	ENSP00000399943	3
AMT	ENST00000430521	?	ENSP00000388068	3
AMT	ENST00000473163	?	?	2
AMT	ENST00000427987	?	ENSP00000403821	3
AMT	ENST00000480957	?	?	1
AMT	ENST00000462048	?	?	1
AMT	ENST00000487589	?	?	1
AMT	ENST00000461210	?	?	1
AMT	ENST00000493046	?	?	1
AMT	ENST00000485108	?	?	1
AMT	ENST00000476828	?	?	1
AMT	ENST00000491800	?	?	1
AMT	ENST00000538581	NM_001164711.1	ENSP00000443200	3
AMT	ENST00000458307	NM_001164710.1	ENSP00000415619	3
AMT	ENST00000476127	?	?	2
AMT	ENST00000476226	?	?	3
AMT	ENST00000478594	?	?	1
AMT	ENST00000495436	?	?	3
AMT	ENST00000273588	NM_000481.3	ENSP00000273588	3
AMT	ENST00000465925	?	?	3
AMT	ENST00000395338	NM_001164712.1	ENSP00000378747	3
APEH	ENST00000296456	NM_001640.3	ENSP00000296456	2
APEH	ENST00000463995	?	?	1
APEH	ENST00000449966	?	ENSP00000414369	1
APEH	ENST00000447436	?	ENSP00000401871	1
APEH	ENST00000457042	?	ENSP00000410366	1
APEH	ENST00000491799	?	?	1
APEH	ENST00000446089	?	ENSP00000410149	1
APEH	ENST00000483715	?	?	1
APEH	ENST00000438011	?	ENSP00000415862	2
APEH	ENST00000442186	?	ENSP00000402365	1
APEH	ENST00000462439	?	?	1
APEH	ENST00000469362	?	ENSP00000438180	1
APEH	ENST00000463616	?	?	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	30

BSN	ENST00000467456	?	?	3
BSN-AS1	ENST00000442384	?	?	2
BSN-AS2	ENST00000421598	?	?	1
BSN-AS2	ENST00000433882	?	?	1
DAG1	ENST00000421560	?	ENSP00000412067	21
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	21
DAG1	ENST00000469139	?	?	17
DAG1	ENST00000461492	?	?	21
DAG1	ENST00000496474	?	?	17
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	21
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	21
DAG1	ENST00000430636	?	ENSP00000401805	17
DAG1	ENST00000466701	?	?	17
DAG1	ENST00000475424	?	?	10
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	21
DAG1	ENST00000419218	?	ENSP00000402443	17
DAG1	ENST00000452060	?	ENSP00000410145	17
DAG1	ENST00000539901	NM_001177644.2	ENSP00000439334	21
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	21
DAG1	ENST00000431960	?	ENSP00000388833	21
DAG1	ENST00000461987	?	?	4
DAG1	ENST00000435508	?	ENSP00000415321	17
DAG1	ENST00000452317	?	ENSP00000387859	17
DAG1	ENST00000479935	?	?	4
DAG1	ENST00000428779	?	ENSP00000401382	17
DAG1	ENST00000418588	?	ENSP00000405859	17
NICN1	ENST00000423832	?	ENSP00000413115	1
NICN1	ENST00000494057	?	?	1
NICN1	ENST00000422593	?	?	1
NICN1	ENST00000497742	?	?	1
NICN1	ENST00000273598	NM_032316.3	ENSP00000273598	1
NICN1	ENST00000436744	?	ENSP00000402335	1
NICN1	ENST00000461015	?	?	1
NICN1	ENST00000615713	?	ENSP00000478287	1
RHOA	ENST00000445425	?	ENSP00000408402	15
RHOA	ENST00000454011	?	ENSP00000394483	16
RHOA	ENST00000422781	?	ENSP00000413587	16
RHOA	ENST00000418115	NM_001664.2	ENSP00000400175	16
RHOA	ENST00000431929	?	?	11
RNF123	ENST00000486102	?	?	2
RNF123	ENST00000454491	?	ENSP00000408490	2

RNF123	ENST00000457726	?	ENSP00000394369	2
RNF123	ENST00000443204	?	ENSP00000390856	2
RNF123	ENST00000487805	?	?	2
RNF123	ENST00000629802	?	ENSP00000487104	2
RNF123	ENST00000327697	NM_022064.3	ENSP00000328287	2
RNF123	ENST00000432042	?	ENSP00000392443	2
TCTA	ENST00000488385	?	?	1
TCTA	ENST00000493381	?	?	1
TCTA	ENST00000273590	NM_022171.2	ENSP00000273590	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
AMT	ENST00000399379	?	ENSP00000399943	1
APEH	ENST00000447436	?	ENSP00000401871	1
APEH	ENST00000446089	?	ENSP00000410149	1
APEH	ENST00000447541	?	ENSP00000404353	1
BSN	ENST00000296452	NM_003458.3	ENSP00000296452	1
DAG1	ENST00000541308	NM_001177638.2, NM_001177641.2, NM_001165928.3	ENSP00000440590	1
DAG1	ENST00000545947	NM_001177634.2	ENSP00000442600	1
DAG1	ENST00000308775	NM_001177636.2, NM_004393.5, NM_001177639.2, NM_001177637.2	ENSP00000312435	1
DAG1	ENST00000538711	NM_001177640.2, NM_001177642.2, NM_001177635.2	ENSP00000438421	1
DAG1	ENST00000539901	NM_001177644.2	ENSP00000439334	1
DAG1	ENST00000515359	NM_001177643.2	ENSP00000440705	1
GPX1	ENST00000419349	NM_201397.1	ENSP00000391316	1
TCTA	ENST00000273590	NM_022171.2	ENSP00000273590	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GPX1	ENST00000620890	?	ENSP00000478837	1
GPX1	ENST00000419783	NM_000581.2	ENSP00000407375	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AMT	ENST00000473163	?	1
AMT	ENST00000476127	?	1
AMT	ENST00000476226	?	2
AMT	ENST00000465925	?	2
APEH	ENST00000482301	?	1
APEH	ENST00000463995	?	1
GPX1	ENST00000496791	?	1
MST1	ENST00000484144	?	1
MST1	ENST00000493836	?	2
MST1	ENST00000479115	?	2
MST1	ENST00000488350	?	2

MST1	ENST00000492329	?	2
TCTA	ENST00000487432	?	1
TCTA	ENST00000497786	?	1
TCTA	ENST00000482193	?	1

Locus 4 (sentinel SNP rs9858542 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.