SNiPAcard

Block annotations

Block info

genomic range	chr6:31,430,060-31,816,059
block size	386,000 bp
variant count	45 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.391 [-3.764 – 1.759]	**gene(s) hit or close-by**	AL662801.1, ATP6V1G2, ATP6V1G2-DDX39B, BAG6, C6orf47, C6orf47-AS1, CLIC1, CSNK2B, CSNK2B-LY6G5B--991, DDX39B, DDX39B-AS1, GPANK1, HCP5, LST1, LTA, LTB, LY6G5B, MCCD1, MICB, MSH5, MSH5-SAPCD1, NCR3, NFKBIL1, PPIAP9, PRRC2A, RPL15P4, SNORA38, SNORD117, SNORD84, TNF, VARS, XXbac-BPG16N22.5, XXbac-BPG181B23.6, Y_RNA, Y_RNA, Y_RNA
phastCons	μ = 0.059 [0 – 0.855]	**eQTL gene(s)**	CDSN, DDR1, HLA-DRB6, LST1, MICA, MICB, NOTCH4, PSORS1C2, RNF5, TNXA, XXbac-BPG181B23.7
GERP++	μ = -0.549 [-6.65 – 4.18]	**potentially regulated gene(s)**	AIF1, AIF1, AIF1, AIF1, AIF1, AIF1, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, ATP6V1G2, C2, C2, C2, C2, C2, C2, C2, C2, C4A, C4A, C4A, C4A, C4A, C6orf48, C6orf48, C6orf48, C6orf48, C6orf48, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CFB, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CLIC1, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CYP21A1P, CYP21A1P, CYP21A1P, CYP21A1P, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DHFRP2, DHFRP2, DHFRP2, DHFRP2, DHFRP2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, EHMT2, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCG27, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LSM2, LSM2, LSM2, LSM2, LSM2, LSM2, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5B, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICA, MICA, MICA, MICA, MICA, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, MSH5, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, NCR3, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, STK19, STK19, STK19, STK19, STK19, STK19, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, XXbac-BPG16N22.5, XXbac-BPG181B23.6, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12, XXbac-BPG248L24.13, XXbac-BPG299F13.15, ZBTB12, ZBTB12, ZBTB12, ZBTB12, ZBTB12
CADD score	μ = 4.116 [0.099 – 15.39]	**disease gene(s)**	NCR3, , , , , , , , , , , , , , , , , , , TNF, , , NFKBIL1, , , C4A, , , , , , , CFB, , , , , , C2, , , , , LTA, HLA-B, CDSN

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HIV-1 control	$<1.00\times10^{-25}$	GWAS Catalog	21051598	1
HIV-1 control	$<1.00\times10^{-11}$	GWAS Catalog	20041166	1
Drug-induced liver injury (flucloxacillin)	$<9.00\times10^{-33}$	GWAS Catalog	19483685	1
AIDS progression	$<3.00\times10^{-19}$	GWAS Catalog	19115949	1
Psoriasis	$<2.00\times10^{-26}$	GWAS Catalog	18369459	1

Disease gene annotation

gene	trait	source DB	source entry/link
NCR3	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	MALARIA, MILD, SUSCEPTIBILITY TO	OMIM	MIM:609148
NFKBIL1	RHEUMATOID ARTHRITIS	OMIM	MIM:180300
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
C4A	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
C4A	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
C4A	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
C4A	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561

	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 4A DEFICIENCY	OMIM	MIM:614380	OMIM
	BLOOD GROUP, CHIDO/RODGERS SYSTEM	OMIM	MIM:614374	OMIM
	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700	OMIM
	COMPLEMENT COMPONENT 4, PARTIAL DEFICIENCY OF	OMIM	MIM:120790	OMIM
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	OMIM
	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	OMIM
	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	OMIM
LTA	LEPROSY, SUSCEPTIBILITY TO, 4	OMIM	MIM:610988	OMIM
LTA	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287	orphanet
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426	orphanet
HLA-B	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C4A	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
C4A	Behcet disease	OrphaNet	OrphaNet:117	orphanet
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	orphanet
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	orphanet
CDSN	Hypotrichosis simplex of the scalp	OrphaNet	OrphaNet:90368	orphanet
CDSN	Generalized peeling skin syndrome type B	OrphaNet	OrphaNet:263553	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MICB	?	ENSG00000204516	muscularis mucosae	1.53×10-8 (p-value)	GTEx Portal V6	44
MICB	?	ENSG00000204516	lung	6.83×10-14 (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	lung	5.56×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transformed fibroblasts	1.64×10-10 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	transformed fibroblasts	1.54×10-6 (p-value)	GTEx Portal V6	13

XXbac-BPG181B23.7	?	ENSG00000272221	tibial artery	3.83×10-7 (p-value)	GTEx Portal V6	20
MICB	?	ENSG00000204516	tibial artery	1.47×10-6 (p-value)	GTEx Portal V6	20
MICB	?	ENSG00000204516	breast	1.70×10-8 (p-value)	GTEx Portal V6	23
MICB	?	ENSG00000204516	blood	1.27×10-22 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	blood	2.73×10-6 (p-value)	GTEx Portal V6	12
MICB	?	ENSG00000204516	thyroid	6.43×10-17 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	thyroid	4.07×10-6 (p-value)	GTEx Portal V6	2
DDR1	?	ENSG00000204580	thyroid	7.87×10-6 (p-value)	GTEx Portal V6	2
HLA-DRB6	?	ENSG00000229391	thyroid	6.94×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	skeletal muscle	4.97×10-9 (p-value)	GTEx Portal V6	44
RNF5	?	ENSG00000204308	skeletal muscle	3.42×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	transverse colon	9.05×10-7 (p-value)	GTEx Portal V6	32
MICB	?	ENSG00000204516	sun exposed skin	4.30×10-11 (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	sun exposed skin	9.25×10-7 (p-value)	GTEx Portal V6	2
XXbac-BPG181B23.7	?	ENSG00000272221	sun exposed skin	7.10×10-6 (p-value)	GTEx Portal V6	1
CDSN	?	ENSG00000204539	sun exposed skin	7.80×10-6 (p-value)	GTEx Portal V6	5
MICB	?	ENSG00000204516	left ventricle	1.04×10-11 (p-value)	GTEx Portal V6	44
MICB	?	ENSG00000204516	subcutaneous adipocytes	1.42×10-13 (p-value)	GTEx Portal V6	44
NOTCH4	?	ENSG00000204301	subcutaneous adipocytes	1.45×10-6 (p-value)	GTEx Portal V6	27
MICB	?	ENSG00000204516	visceral adipocytes	5.67×10-10 (p-value)	GTEx Portal V6	35
MICB	?	ENSG00000204516	tibial nerve	3.67×10-10 (p-value)	GTEx Portal V6	44
MICA	?	ENSG00000204520	tibial nerve	7.32×10-6 (p-value)	GTEx Portal V6	1
LST1	?	ENSG00000204482	tibial nerve	3.11×10-6 (p-value)	GTEx Portal V6	1
MICB	?	ENSG00000204516	esophagus mucosa	5.79×10-6 (p-value)	GTEx Portal V6	15
XXbac-BPG181B23.7	?	ENSG00000272221	esophagus mucosa	3.48×10-6 (p-value)	GTEx Portal V6	1
TNXA	?	ENSG00000248290	adrenal gland	1.58×10-8 (p-value)	GTEx Portal V6	44
XXbac-BPG181B23.7	?	ENSG00000272221	stomach	2.95×10-6 (p-value)	GTEx Portal V6	2
MICB	?	ENSG00000204516	blood	1.98×10-3 (q-value)	SeeQTL DB (HapMap)	2
PSORS1C2	ENST00000259845	ILMN_1653447	monocyte	2.10×10-5 (p-value)	Fairfax et al.	4
MICB	ENST00000399150	ILMN_1708006	monocyte	2.79×10-14 (p-value)	Fairfax et al.	4
MICB	ENST00000538442		b-cell	5.86×10-33 (p-value)	Fairfax et al.	4
MICB	ENST00000252229					

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)

ENCP00000048021

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444839	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048018	XXbac-BPG181B23.6
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048107	VARS VARS VARS VARS VARS VARS VARS
		ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048038	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
		ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000047994	XXbac-BPG299F13.15
		ENCP00000048017	HCP5 HCP5 HCP5 HCP5
		ENCP00000048050	PRRC2A PRRC2A

	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048039	NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3 NCR3
ENCP00000047996	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000047991	HCG27 HCG27 HCG27 HCG27 HCG27 HCG27 HCG27
ENCP00000048047	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048066	LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B LY6G5B
ENCP00000048046	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048045	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048109	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048033	TNF

			TNF TNF TNF TNF TNF TNF TNF
		ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048031	ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2 ATP6V1G2
		ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
		ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048006	XXbac-BPG248L24.12
		ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCE00000445135	1	ENCP00000048115	C6orf48 C6orf48 C6orf48 C6orf48 C6orf48
		ENCP00000048132	C2 C2 C2 C2 C2 C2 C2 C2
		ENCP00000048070	LY6G5C LY6G5C

	LY6G5C
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ENCP00000048140	CFB
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ENCP00000048090	CLIC1
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ENCP00000048135	C2
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ENCP00000048111	LSM2
	LSM2
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ENCP00000048130	ZBTB12
	ZBTB12
	ZBTB12
	ZBTB12
	ZBTB12
ENCP00000048102	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
	VARS
ENCP00000048123	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
	EHMT2
ENCP00000048156	C4A
	C4A
	C4A
	C4A
	C4A
ENCP00000048092	CLIC1
	CLIC1
	CLIC1
	CLIC1
	MSH5
	CLIC1
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	MSH5
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			MSH5
		ENCP00000048103	VARS VARS VARS VARS VARS VARS VARS
ENCE00000444818	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048001	XXbac-BPG248L24.13
		ENCP00000048008	DHFRP2 DHFRP2 DHFRP2 DHFRP2 DHFRP2
		ENCP00000048041	AIF1 AIF1 AIF1 AIF1 AIF1 AIF1
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048013	HCP5 MICA MICA HCP5 MICA HCP5 MICA MICA HCP5
		ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
		ENCP00000048017	HCP5 HCP5 HCP5 HCP5
		ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
		ENCP00000048023	DDX39B

	DDX39B
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	DDX39B
ENCP00000047991	HCG27
	HCG27
	HCG27
	HCG27
	HCG27
	HCG27
	HCG27
ENCP00000048104	VARS
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	PRRC2A
ENCP00000048110	VARS
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ENCP00000048109	VARS
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ENCP00000048051	PRRC2A
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ENCP00000047998	HLA-C
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ENCP00000048035	LST1
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ENCP00000048043	AIF1
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ENCP00000048063	GPANK1
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ENCP00000048048	PRRC2A
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		ENCP00000048040	UQCRHP1
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ENCP00000048003	HLA-B
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ENCP00000048007	HLA-B
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ENCP00000048048	PRRC2A
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ENCP00000048093	MSH5
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ENCP00000048134	C2
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ENCP00000048037	LST1
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ENCP00000048020	MICB
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	GPANK1
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	CSNK2B
	GPANK1
	GPANK1
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ENCP00000048069	LY6G5C
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ENCP00000048005	HLA-B
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ENCP00000048017	HCP5
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ENCP00000048023	DDX39B
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	ENCP00000048052	PRRC2A
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	ENCP00000048157	CYP21A1P
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		CYP21A1P
		CYP21A1P
	ENCP00000048063	GPANK1
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		GPANK1
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		GPANK1
		GPANK1
	ENCP00000048040	UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1
		UQCRHP1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000129758 (promoter flanking region)	2	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, DNase1, TAF7, PolII, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K4me2, H2AZ, H3K36me3
		blood (K562)	H3K27me3, Cfos
		skin (NHDF-AD)	DNase1, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, PolII, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me3, PolII, H3K4me2, H3K27me3, DNase1
		lung (IMR90)	H3K27me3
		blood (GM12878)	TAF1, PolII, H2AZ, DNase1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K4me2, H3K9ac

		A549	DNase1, H3K4me3
		skin (NHEK)	H3K4me3, DNase1, H3K4me2, H3K9ac
ENSR00001703053 (enhancer)	1	Osteobl	H3K4me2
		nervous (NH-A)	H3K27me3
		blood (K562)	H3K27me3
ENSR00000264405 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, Nrsf, CTCF, USF1, DNase1
		HSMMtube	CTCF
		Osteobl	CTCF
		blood (K562)	CTCF, Nrsf, PolII, Max, Rad21, USF1
		blood (DND-41)	CTCF
		skin (NHDF-AD)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	DNase1, CTCF
		endothelium (HUVEC)	Max, CTCF
		liver (HepG2)	Nrsf, Rad21, USF1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	CTCF
ENSR00000264406 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, H3K36me3, H3K27me3, PolII, Rad21, TAF7, CTCF, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	CTCF, H3K4me2, DNase1, H3K27me3, H2AZ
		blood (K562)	Max, ZBTB7A, Rad21, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, CTCF, DNase1, H3K4me3, Ini1, H3K27me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, H3K79me2, CTCF, H3K4me3, H3K4me2
		liver (HepG2)	DNase1, H3K27me3, PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, CTCF, H3K4me3
		lung (IMR90)	H3K4me2, H3K4me3, CTCF
		blood (GM12878)	PolII, H2AZ, DNase1, Rad21, H3K79me2, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	H3K9ac, CTCF, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	H3K9ac, H4K20me1, H3K4me3, H3K27ac, H3K4me2, DNase1, H3K4me1, CTCF
		endothelium (HUVEC)	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000487848 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, PolII, TAF7, TAF1, H3K4me2, USF1, H3K4me3, DNase1
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	Max, ZBTB7A, CTCF, H3K9ac, H2AZ, TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	PolII, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4me2
		blood (GM12878)	PolII, H2AZ, DNase1, CTCF, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	H3K4me2, H3K4me3, DNase1, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H2AZ, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K9ac, H3K27ac, CTCF, H3K4me1
		breast (HMEC)	H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, PolII, DNase1
		monocytes (Monocytes-CD14+)	H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K4me3
		endothelium (HUVEC)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, H3K36me3
		A549	H3K4me3, H3K4me2, H3K9ac
ENSR00001494568 (promoter flanking region)	2	liver (HepG2)	H3K27me3
		HSMMtube	DNase1
		blood (K562)	H3K27me3
ENSR00000487851 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K4me1
		embryonic stem cell (H1ESC)	H3K27me3, Rad21
		liver (HepG2)	H3K27me3

		blood (DND-41)	H3K27me3
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1
ENSR00000487856 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, Nrsf, Sin3Ak20, H3K27ac, Yy1, HDAC2, TAF1, H3K4me2, SP1, H3K36me3, H3K9ac, H3K4me3, TAF7, Gabp, H4K5ac, H3K27me3, CTCF, PolII
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	Yy1, Ini1, Brg1, THAP1, H3K27ac, Max, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Nrsf, Cmyc, PolII, H4K20me1, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K27me3, H3K36me3, H3K4me3, Cmyc, Gabp, Yy1, Nrsf, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	DNase1, H4K8ac, H3K14ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac, H3K56ac
		blood (GM12878)	TAF1, Nrsf, PolII, H2AZ, DNase1, Cmyc, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3
		nervous (NH-A)	DNase1, H3K36me3, H3K9ac, H4K20me1, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	H3K36me3, H3K4me3, H3K4me2, DNase1, H3K4me1, H3K27ac, H4K20me1, H3K9ac, PolII
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H4K20me1, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H4K20me1, H3K9ac, H3K4me3, H3K27me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K9ac, Nrsf, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc, H3K36me3
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000487858 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, Rad21, H3K36me3
		HSMMtube	CTCF
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF, H3K36me3
		liver (HepG2)	H3K36me3, Rad21, CTCF
		lung (IMR90)	H3K36me3, CTCF
		blood (GM12878)	CTCF, Rad21
		skin (NHEK)	CTCF, H3K36me3
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF
		A549	H3K36me3
ENSR00000487863 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		blood (K562)	H3K4me1, PU1, FOSL1, H3K27me3, PolII, Ini1, Brg1, DNase1
		muscle (HSMM)	DNase1
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K4me1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		blood (GM12878)	H3K4me1, H3K27ac
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
ENSR00001494573 (promoter)	1	NHLF	H3K27me3
		embryonic stem cell (H1ESC)	DNase1, H3K27me3
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1, H3K27ac, H3K9ac, H3K27me3, H3K36me3
		blood (K562)	H3K27me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K27me3
		blood (GM12878)	EBF1, POU2F2, Cmyc, PolII, DNase1, Egr1, Yy1, Gabp, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3

		lung (IMR90)	H3K27me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K4me2
ENSR00000487869 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, Nrsf
		blood (K562)	H3K27me3
		skin (NHDF-AD)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K36me3, H4K20me1
		liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K36me3
		skin (NHEK)	DNase1
ENSR00000264430 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, Yy1, TAF1, H3K4me2, SP1, USF1, H3K36me3, H3K9ac, H3K4me3, SIX5, Sin3Ak20, H4K5ac, H3K27me3, CTCF, PolII, TAF7
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	Ini1, H2AZ, TAF1, ELF1, Cjun, PolII, GTF2B, H3K4me2, H3K36me3, DNase1, H3K4me3, HEY1, H3K79me2, Brg1, Egr1, FOSL1, SIX5, H3K27ac, Max, TAF7, H3K4me1, E2F6, H3K9ac, Gabp, Jund, ETS1, USF1, Cmyc, H3K27me3, Yy1
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		liver (HepG2)	DNase1, Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, USF1, H3K4me1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3
		blood (GM12878)	H3K9ac, H3K36me3, H3K4me2, H3K27ac, Tr4, SIX5, PolII, Srf, H2AZ, ETS1, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, H3K4me2, H3K27ac, DNase1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K36me3, DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cjun, Max, H3K4me3, H3K4me2, PolII, DNase1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3
ENSR00000487898 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H3K4me3, PolII, Rad21, Jund, Nrsf, Sin3Ak20, CTCF, H3K27ac, ATF3, Yy1, H3K4me1, TAF1, H3K4me2, SP1, USF1, H3K9ac
		HSMMtube	H3K9ac, DNase1, H3K36me3, H2AZ, H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K4me3, Jund, H3K79me2, HEY1, H2AZ, TAF1, ELF1, Cjun, Cfos, Cmyc, PolII, H4K20me1, H3K4me2, CTCF, H3K4me1, H3K36me3, H3K9ac, E2F6, Yy1, Ini1, Brg1, Egr1, FOSL1, H3K27ac, Max, SP2, ZBTB7A, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me2, HNF4A, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, H3K36me3, p300, H3K4me1, FOSL2, Jund, HNF4G, FOXA2, PolII, HDAC2, H3K79me2, TAF1, RXRA, ELF1
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3
		blood (GM12878)	H3K27ac, Cfos, H3K4me2, H3K9ac, H3K36me3, H3K4me3, H3K79me2, PolII, H2AZ, H3K4me1, DNase1, CTCF, Yy1, ELF1
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3
		skin (NHEK)	H3K4me2, H3K4me3, H3K36me3, DNase1, H3K9ac, H3K27ac, H3K4me1, CTCF
		NHLF	H3K27ac, H3K36me3, DNase1, H3K4me3, H3K9ac
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K4me2, CTCF, H3K27ac, H3K9ac, H3K4me3, H3K36me3, H3K4me1
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3, CTCF
		cervix (HeLa-S3)	DNase1, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, H3K79me2, Gabp, PolII, Cmyc, CTCF, H3K36me3, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, Cmyc, CTCF, PolII, H3K27ac, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K4me1
		A549	H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2, H3K4me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
HCP5	1	ENST00000414046 ENST00000467369 ENST00000541196	hsa-miR-181a-5p hsa-miR-181b-5p hsa-miR-181d-5p hsa-miR-22-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL662801.1	upstream gene variant	2780	ENST00000617927	?	?	1
ATP6V1G2	downstream gene variant, upstream gene variant	1436	ENST00000483251	NM_138282.2	ENSP00000419698	3
ATP6V1G2	downstream gene variant, upstream gene variant	1482	ENST00000483170	?	?	3
ATP6V1G2	downstream gene variant, upstream gene variant	1474	ENST00000415099	?	ENSP00000390148	4
ATP6V1G2	downstream gene variant, upstream gene variant	660	ENST00000303892	NM_130463.3	ENSP00000302194	3
ATP6V1G2	downstream gene variant, upstream gene variant	1954	ENST00000481998	?	?	3
ATP6V1G2	downstream gene variant, upstream gene variant	660	ENST00000376151	NM_001204078.1	ENSP00000365321	3
ATP6V1G2-DDX39B	downstream gene variant, upstream gene variant	4765	ENST00000480131	?	ENSP00000420191	2
ATP6V1G2-DDX39B	upstream gene variant	4995	ENST00000475917	?	?	1
BAG6	downstream gene variant, upstream gene variant	1459	ENST00000469182	?	?	2
BAG6	downstream gene variant, upstream gene variant	1567	ENST00000462875	?	?	3
BAG6	downstream gene variant, upstream gene variant	1951	ENST00000438149	?	ENSP00000410280	2
BAG6	downstream gene variant, upstream gene variant	598	ENST00000465348	?	?	3
BAG6	downstream gene variant	768	ENST00000456286	?	ENSP00000406436	2
BAG6	downstream gene variant	573	ENST00000428326	?	ENSP00000413561	2
BAG6	upstream gene variant	1835	ENST00000462682	?	?	1
BAG6	upstream gene variant	4259	ENST00000464126	?	?	1
BAG6	downstream gene variant	660	ENST00000424176	?	ENSP00000391783	2
BAG6	downstream gene variant	571	ENST00000456622	?	ENSP00000401031	2
BAG6	downstream gene variant	571	ENST00000434444	?	ENSP00000394856	2
BAG6	downstream gene variant	1892	ENST00000454165	?	ENSP00000396503	2
BAG6	downstream gene variant	584	ENST00000424480	?	ENSP00000411043	2
BAG6	upstream gene variant	3861	ENST00000422948	?	ENSP00000413225	1
BAG6	downstream gene variant	574	ENST00000451898	?	ENSP00000391613	2
BAG6	downstream gene variant	580	ENST00000433828	?	ENSP00000388244	2
BAG6	downstream gene variant	2961	ENST00000437771	?	ENSP00000397978	1
BAG6	downstream gene variant	571	ENST00000452994	?	ENSP00000404971	2
BAG6	downstream gene variant, upstream gene variant	1517	ENST00000453833	?	ENSP00000406434	2
BAG6	downstream gene variant	686	ENST00000441054	?	ENSP00000406302	2
BAG6	upstream gene variant	3570	ENST00000441793	?	ENSP00000416633	1
BAG6	downstream gene variant, upstream gene variant	482	ENST00000470875	?	?	3
C6orf47	upstream gene variant	4933	ENST00000375911	NM_021184.3	ENSP00000365076	1
C6orf47-AS1	downstream gene variant	4984	ENST00000422049	?	?	1
CLIC1	upstream gene variant	3182	ENST00000375784	NM_001288.4	ENSP00000364940	1
CLIC1	upstream gene variant	2431	ENST00000375780	NM_001287593.1	ENSP00000364935	1

CLIC1	upstream gene variant	3204	ENST00000616760	NM_001287594.1	ENSP00000479808	1
CLIC1	upstream gene variant	3177	ENST00000375779	?	ENSP00000364934	1
CSNK2B	upstream gene variant	382	ENST00000465481	?	?	1
CSNK2B	upstream gene variant	359	ENST00000375882	NM_001320.6, NM_001282385.1	ENSP00000365042	1
CSNK2B	upstream gene variant	389	ENST00000481269	?	?	1
CSNK2B	upstream gene variant	376	ENST00000468255	?	?	1
CSNK2B	upstream gene variant	396	ENST00000475875	?	?	1
CSNK2B	upstream gene variant	650	ENST00000375866	?	ENSP00000365026	1
CSNK2B	upstream gene variant	587	ENST00000375865	?	ENSP00000365025	1
CSNK2B-LY6G5B--991	upstream gene variant	175	ENST00000618754	?	ENSP00000478610	1
CSNK2B-LY6G5B--991	upstream gene variant	175	ENST00000622499	?	ENSP00000483024	1
CSNK2B-LY6G5B--991	upstream gene variant	397	ENST00000375880	?	ENSP00000365040	1
CSNK2B-LY6G5B--991	upstream gene variant	3413	ENST00000409691	?	ENSP00000387322	1
CSNK2B-LY6G5B--991	upstream gene variant	1115	ENST00000617558	?	ENSP00000483989	1
DDX39B	upstream gene variant	719	ENST00000419338	?	ENSP00000410313	2
DDX39B	upstream gene variant	1354	ENST00000458640	NM_080598.5	ENSP00000416269	1
DDX39B	upstream gene variant	185	ENST00000428098	?	ENSP00000392672	2
DDX39B	upstream gene variant	1354	ENST00000396172	NM_004640.6	ENSP00000379475	1
DDX39B	upstream gene variant	1138	ENST00000417023	?	ENSP00000406422	1
DDX39B	upstream gene variant	174	ENST00000484566	?	ENSP00000436220	1
DDX39B	upstream gene variant	3905	ENST00000474961	?	?	1
DDX39B	upstream gene variant	1667	ENST00000376177	?	ENSP00000365347	2
DDX39B	upstream gene variant	4714	ENST00000462421	?	?	1
DDX39B	downstream gene variant, upstream gene variant	200	ENST00000419020	?	ENSP00000405245	3
DDX39B	upstream gene variant	3978	ENST00000478365	?	?	1
DDX39B	upstream gene variant	202	ENST00000481456	?	?	2
DDX39B	downstream gene variant, upstream gene variant	212	ENST00000418897	?	ENSP00000399841	3
DDX39B	upstream gene variant	222	ENST00000431908	?	ENSP00000408000	2
DDX39B	downstream gene variant, upstream gene variant	214	ENST00000428450	?	ENSP00000405707	3
DDX39B	upstream gene variant	672	ENST00000427214	?	ENSP00000399371	2
DDX39B	downstream gene variant, upstream gene variant	217	ENST00000456976	?	ENSP00000393984	3
DDX39B	upstream gene variant	266	ENST00000456662	?	ENSP00000416350	2
DDX39B	downstream gene variant, upstream gene variant	474	ENST00000449757	?	ENSP00000409426	3
DDX39B	upstream gene variant	186	ENST00000462256	?	?	2
DDX39B	upstream gene variant	1401	ENST00000482195	?	?	1
DDX39B-AS1	upstream gene variant, downstream gene variant	170	ENST00000416684	?	?	2
DDX39B-AS1	upstream gene variant, downstream	101	ENST00000420520	?	?	2

	gene variant					
GPANK1	upstream gene variant	315	ENST00000375896	NM_033177.3	ENSP00000365060	1
GPANK1	upstream gene variant	75	ENST00000375893	NM_001199240.1	ENSP00000365057	1
GPANK1	upstream gene variant	263	ENST00000458083	?	ENSP00000395307	1
GPANK1	upstream gene variant	515	ENST00000445768	?	ENSP00000409349	1
GPANK1	upstream gene variant	520	ENST00000456540	?	ENSP00000395484	1
HCP5	upstream gene variant, downstream gene variant	488	ENST00000467369	?	?	2
HCP5	upstream gene variant, downstream gene variant	182	ENST00000460889	?	?	2
HCP5	upstream gene variant	899	ENST00000541196	?	?	1
LST1	downstream gene variant	1825	ENST00000376092	?	ENSP00000365260	1
LST1	downstream gene variant	1578	ENST00000376089	?	ENSP00000365257	1
LST1	downstream gene variant	1578	ENST00000376093	NM_007161.3	ENSP00000365261	1
LST1	downstream gene variant	1578	ENST00000418507	NM_205838.2	ENSP00000405900	1
LST1	downstream gene variant	698	ENST00000433492	?	ENSP00000399869	2
LST1	downstream gene variant	1578	ENST00000396101	?	ENSP00000379408	1
LST1	downstream gene variant	1578	ENST00000303757	NM_205840.2	ENSP00000303649	1
LST1	downstream gene variant	1578	ENST00000376090	?	ENSP00000365258	1
LST1	downstream gene variant	1578	ENST00000376099	?	ENSP00000365267	1
LST1	downstream gene variant	1578	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	downstream gene variant	587	ENST00000419073	?	?	2
LST1	downstream gene variant	1811	ENST00000490742	?	ENSP00000418578	1
LST1	downstream gene variant	1730	ENST00000464044	?	ENSP00000417645	1
LST1	downstream gene variant	1730	ENST00000376110	?	ENSP00000365278	1
LST1	downstream gene variant	1579	ENST00000376086	NM_001166538.1	ENSP00000365254	1
LST1	downstream gene variant	1677	ENST00000211921	?	ENSP00000211921	1
LST1	downstream gene variant	1578	ENST00000464526	?	?	1
LST1	downstream gene variant	1861	ENST00000396112	?	ENSP00000379418	1
LST1	downstream gene variant	1677	ENST00000339530	NM_205837.2	ENSP00000339201	1
LST1	downstream gene variant	1578	ENST00000376096	?	ENSP00000365264	1
LST1	downstream gene variant	1730	ENST00000460834	?	?	1
LTA	downstream gene variant	1657	ENST00000489638	?	?	3
LTA	downstream gene variant	1657	ENST00000454783	NM_001159740.2	ENSP00000403495	3
LTA	downstream gene variant	1657	ENST00000418386	NM_000595.3	ENSP00000413450	3
LTA	downstream gene variant	1657	ENST00000471842	?	?	3
LTB	downstream gene variant	1187	ENST00000483972	?	?	4
LTB	downstream gene variant	1217	ENST00000429299	NM_002341.1	ENSP00000410481	4
LTB	downstream gene variant	1217	ENST00000446745	NM_009588.1	ENSP00000416113	4
LTB	downstream gene variant	1286	ENST00000482429	?	?	4

LY6G5B	upstream gene variant	4674	ENST00000409525	?	ENSP00000386365	1
LY6G5B	upstream gene variant	4462	ENST00000375864	NM_021221.2	ENSP00000365024	1
MCCD1	upstream gene variant	3624	ENST00000376191	NM_001011700.2	ENSP00000365362	2
MICB	upstream gene variant, downstream gene variant	59	ENST00000399150	NM_001289161.1	ENSP00000382103	10
MICB	upstream gene variant, downstream gene variant	10	ENST00000538442	NM_001289160.1	ENSP00000442345	10
MICB	upstream gene variant, downstream gene variant	59	ENST00000252229	NM_005931.4	ENSP00000252229	10
MICB	downstream gene variant	320	ENST00000494577	?	?	5
MSH5	upstream gene variant, downstream gene variant	242	ENST00000425703	?	ENSP00000402842	2
MSH5	upstream gene variant	256	ENST00000482280	?	?	1
MSH5	upstream gene variant	202	ENST00000375750	NM_172166.3	ENSP00000364903	1
MSH5	upstream gene variant	4229	ENST00000450148	?	ENSP00000394971	1
MSH5	upstream gene variant	477	ENST00000423982	?	ENSP00000406352	1
MSH5	upstream gene variant	199	ENST00000375755	NM_002441.4	ENSP00000364908	1
MSH5	upstream gene variant	1497	ENST00000463144	?	ENSP00000419648	1
MSH5	upstream gene variant	272	ENST00000375703	NM_172165.3	ENSP00000364855	1
MSH5	upstream gene variant	272	ENST00000375740	NM_025259.5	ENSP00000364892	1
MSH5	upstream gene variant, downstream gene variant	256	ENST00000463094	?	?	2
MSH5	upstream gene variant	4482	ENST00000497269	?	ENSP00000419131	1
MSH5-SAPCD1	upstream gene variant	271	ENST00000493662	?	ENSP00000417871	1
NCR3	downstream gene variant, upstream gene variant	467	ENST00000491161	?	?	2
NCR3	downstream gene variant, upstream gene variant	846	ENST00000376072	NM_001145467.1	ENSP00000365240	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000376073	NM_001145466.1	ENSP00000365241	2
NCR3	downstream gene variant, upstream gene variant	869	ENST00000376071	?	ENSP00000365239	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000340027	NM_147130.2	ENSP00000342156	2
NCR3	downstream gene variant, upstream gene variant	467	ENST00000495600	?	?	2
NFKBIL1	upstream gene variant, downstream gene variant	3779	ENST00000376148	NM_005007.3	ENSP00000365318	2
NFKBIL1	upstream gene variant, downstream gene variant	3811	ENST00000376145	NM_001144961.1	ENSP00000365315	2
NFKBIL1	upstream gene variant, downstream gene variant	3068	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	3
NFKBIL1	upstream gene variant	3859	ENST00000473655	?	?	1
NFKBIL1	upstream gene variant	3838	ENST00000496233	?	ENSP00000437148	1
PPIAP9	downstream gene variant, upstream gene variant	3365	ENST00000403866	?	?	4
PRRC2A	downstream gene variant	3549	ENST00000469501	?	?	1
PRRC2A	downstream gene variant	3008	ENST00000487089	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000376007	NM_080686.2	ENSP00000365175	1
PRRC2A	downstream gene variant	1487	ENST00000492691	?	?	1
PRRC2A	downstream gene variant	3942	ENST00000484787	?	?	1
PRRC2A	downstream gene variant	4502	ENST00000460302	?	?	1

PRRC2A	downstream gene variant	2447	ENST00000487839	?	?	1
PRRC2A	upstream gene variant	34	ENST00000464079	?	?	1
PRRC2A	upstream gene variant	886	ENST00000464890	?	?	1
PRRC2A	upstream gene variant	3772	ENST00000483470	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000462617	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000482441	?	?	1
PRRC2A	downstream gene variant	1487	ENST00000376033	NM_004638.3	ENSP00000365201	1
RPL15P4	upstream gene variant	3021	ENST00000416625	?	?	2
SNORA38	downstream gene variant	1658	ENST00000363946	?	?	1
SNORD117	downstream gene variant	725	ENST00000364915	?	?	1
SNORD84	upstream gene variant	1025	ENST00000584275	?	?	2
TNF	downstream gene variant	382	ENST00000449264	NM_000594.3	ENSP00000398698	3
VARS	upstream gene variant	3243	ENST00000463184	?	?	1
VARS	upstream gene variant	705	ENST00000428445	?	ENSP00000415679	1
VARS	upstream gene variant	874	ENST00000461874	?	?	1
VARS	upstream gene variant	55	ENST00000474643	?	?	1
VARS	upstream gene variant	1100	ENST00000482996	?	?	1
VARS	downstream gene variant	1304	ENST00000495010	?	?	1
VARS	upstream gene variant	1544	ENST00000459667	?	?	1
VARS	downstream gene variant	1952	ENST00000489979	?	?	1
VARS	upstream gene variant	865	ENST00000483275	?	?	1
VARS	upstream gene variant	1369	ENST00000479051	?	?	1
XXbac-BPG16N22.5	upstream gene variant	1130	ENST00000538358	?	?	7
XXbac-BPG181B23.6	downstream gene variant, upstream gene variant	445	ENST00000440087	?	?	2
Y_RNA	upstream gene variant	4389	ENST00000364685	?	?	1
Y_RNA	downstream gene variant, upstream gene variant	375	ENST00000383850	?	?	4
Y_RNA	downstream gene variant	2304	ENST00000364337	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ATP6V1G2-DDX39B	ENST00000376185	?	ENSP00000365356	3
ATP6V1G2-DDX39B	ENST00000475917	?	?	2
ATP6V1G2-DDX39B	ENST00000480131	?	ENSP00000420191	2
BAG6	ENST00000437771	?	ENSP00000397978	2
BAG6	ENST00000464126	?	?	1
BAG6	ENST00000439687	NM_001199697.1	ENSP00000402856	3
		NM_080703.2, NM_080702.2		3

BAG6	ENST00000211379		ENSP00000211379	
BAG6	ENST00000462682	?	?	1
BAG6	ENST00000375976	NM_001098534.1	ENSP00000365143	3
BAG6	ENST00000464869	?	?	1
BAG6	ENST00000362049	NM_001199698.1	ENSP00000354875	3
BAG6	ENST00000469182	?	?	1
BAG6	ENST00000438149	?	ENSP00000410280	1
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DDX39B	ENST00000456662	?	ENSP00000416350	1
DDX39B	ENST00000376177	?	ENSP00000365347	1
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LST1	ENST00000438075	NM_205839.2	ENSP00000391929	1
LST1	ENST00000376092	?	ENSP00000365260	1
LST1	ENST00000303757	NM_205840.2	ENSP00000303649	1
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MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	3
MICB	ENST00000252229	NM_005931.4	ENSP00000252229	3
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MSH5	ENST00000375755	NM_002441.4	ENSP00000364908	1
MSH5	ENST00000450148	?	ENSP00000394971	1
MSH5	ENST00000375750	NM_172166.3	ENSP00000364903	1
MSH5	ENST00000482280	?	?	1
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NFKBIL1	ENST00000376146	NM_001144962.1, NM_001144963.1	ENSP00000365316	2
NFKBIL1	ENST00000496233	?	ENSP00000437148	2
PRRC2A	ENST00000376033	NM_004638.3	ENSP00000365201	1
PRRC2A	ENST00000376007	NM_080686.2	ENSP00000365175	1
PRRC2A	ENST00000469577	?	?	1
TNF	ENST00000449264	NM_000594.3	ENSP00000398698	1
VARS	ENST00000375663	NM_006295.2	ENSP00000364815	1
VARS	ENST00000461328	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CLIC1	ENST00000395892	?	ENSP00000379229	1
DDX39B	ENST00000396172	NM_004640.6	ENSP00000379475	1
DDX39B	ENST00000458640	NM_080598.5	ENSP00000416269	1
GPANK1	ENST00000375900	NM_001199238.1, NM_001199239.1	ENSP00000365065	1
GPANK1	ENST00000375895	?	ENSP00000365059	1
MICB	ENST00000399150	NM_001289161.1	ENSP00000382103	1

MICB	ENST00000252229	NM_005931.4	ENSP00000252229	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ATP6V1G2-DDX39B	ENST00000475917	?	1
DDX39B	ENST00000482195	?	1
HCP5	ENST00000414046	?	1
HCP5	ENST00000541196	?	1
LST1	ENST00000464526	?	1
XXbac-BPG16N22.5	ENST00000538358	?	1