Locus 199 (rs3129682)

SNiPAcard

Block annotations

Block info

genomic range	chr6:28,787,384-29,523,957
block size	736,574 bp
variant count	158 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.194 [-5.844 – 4.77]	**gene(s) hit or close-by**	AL645937.1, AL662791.1, GABBR1, HCG14, HCG15, HCG16, KRT18P1, LINC01556, MAS1L, MAS1LP1, OR10C1, OR10C1, OR11A1, OR12D3, OR14J1, OR2AD1P, OR2B3, OR2B4P, OR2H1, OR2H4P, OR2I1P, OR2J2, OR2J3, OR2J4P, OR2N1P, OR2P1P, OR2U2P, OR2W1, OR5V1, RN7SL471P, RNU6-930P, RPL13P, RPS17P1, SAR1P1, TRIM27, UBD, UBDP1, XXbac-BCX196D17.5, XXbac-BPG13B8.10, XXbac-BPG13B8.11, XXbac-BPG258E24.10, XXbac-BPG308J9.3, XXbac-BPG308K3.5, XXbac-BPG308K3.6, ZNF311, ZNF90P2
phastCons	μ = 0.110 [0 – 1]	**eQTL gene(s)**	ABT1, AL022393.7, BTN3A2, BTN3A3, C2, CFB, GABBR1, HIST1H3J, HIST1H4J, HLA-A, HLA-C, HLA-F-AS1, HLA-G, HLA-H, HLA-J, HLA-K, HLA-L, HLA-U, HLA-V, OR2B7P, OR2B8P, RP1-265C24.5, RP1-313I6.12, SLC44A4, TRIM31, XXbac-BPG308K3.5, ZDHHC20P1, ZFP57, ZNF165, ZNF192P1, ZNRD1, ZNRD1-AS1_3, ZSCAN23, ZSCAN26, ZSCAN31
GERP++	μ = -0.271 [-5.5 – 2.32]	**potentially regulated gene(s)**	AL662791.1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, DDX39BP1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GABBR1, GPX5, GPX5, GPX6, GPX6, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, HLA-H, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR12D3, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2AD1P, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B3, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2B4P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2G1P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2H4P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2N1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2P1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, OR2U1P, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, TRIM27, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5, XXbac-BCX196D17.5
CADD score	μ = 4.022 [0.006 – 14.6]	**disease gene(s)**	ZFP57, TRIM27, OR2J3, , , , , C2, CFB, HLA-A

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Barrett's esophagus	$<4.00\times10^{-9}$	GWAS Catalog	22961001	1
phenol sulfate	2.11×10^{-6}	Metabolomics GWAS Server	24816252	8
betaine	4.49×10^{-7}	Metabolomics GWAS Server	24816252	20
Lupus erythematosus, systemic	9.64×10^{-6}	dbGaP	pha002867	4
Lupus erythematosus, systemic	8.06×10^{-9}	dbGaP	pha002848	1

Disease gene annotation

gene	trait	source DB	source entry/link
ZFP57	DIABETES MELLITUS, TRANSIENT NEONATAL, 1	OMIM	MIM:601410
TRIM27	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550
OR2J3	C3HEX, ABILITY TO SMELL	OMIM	MIM:615082

	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
	THYROID CARCINOMA, PAPILLARY	OMIM	MIM:188550	
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000	
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	
CFB	COMPLEMENT FACTOR B DEFICIENCY	OMIM	MIM:615561	
CFB	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, 4	OMIM	MIM:612924	
CFB	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075	
ZFP57	Diabetes Mellitus, 6q24-Related Transient Neonatal	DECIPHER	MIM:601410	
ZFP57	Transient neonatal diabetes mellitus	OrphaNet	OrphaNet:99886	
TRIM27	Papillary or follicular thyroid carcinoma	OrphaNet	OrphaNet:146	
HLA-A	Birdshot chorioretinopathy	OrphaNet	OrphaNet:179	
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147	
CFB	Atypical hemolytic uremic syndrome with B factor anomaly	OrphaNet	OrphaNet:93578	

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)
OR12D3	missense variant	ENST00000396806	NM_030959.2	ENSP00000380023	I/T	aTc/aCc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ZFP57	?	ENSG00000204644	muscularis mucosae	4.17×10-8 (p-value)	GTEx Portal V6	153
AL022393.7	?	ENSG00000216901	muscularis mucosae	6.78×10-6 (p-value)	GTEx Portal V6	5
HLA-H	?	ENSG00000206341	muscularis mucosae	3.17×10-6 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	lung	1.19×10-9 (p-value)	GTEx Portal V6	155
HLA-G	?	ENSG00000204632	lung	1.18×10-6 (p-value)	GTEx Portal V6	81
HLA-H	?	ENSG00000206341	lung	2.28×10-7 (p-value)	GTEx Portal V6	98
HLA-J	?	ENSG00000204622	lung	9.19×10-10 (p-value)	GTEx Portal V6	67
ZFP57	?	ENSG00000204644	atrial appendage	2.14×10-9 (p-value)	GTEx Portal V6	126
ZSCAN26	?	ENSG00000197062	transformed fibroblasts	7.66×10-6 (p-value)	GTEx Portal V6	108
ZFP57	?	ENSG00000204644	transformed fibroblasts	1.02×10-7 (p-value)	GTEx Portal V6	134
ZFP57	?	ENSG00000204644	blood	2.31×10-12 (p-value)	GTEx Portal V6	155
HLA-F-AS1	?	ENSG00000214922	blood	1.81×10-7 (p-value)	GTEx Portal V6	101
HLA-H	?	ENSG00000206341	blood	9.34×10-8 (p-value)	GTEx Portal V6	84
HLA-K	?	ENSG00000230795	blood	8.61×10-9 (p-value)	GTEx Portal V6	28
HLA-J	?	ENSG00000204622	blood	5.11×10-7 (p-value)	GTEx Portal V6	19
ZFP57	?	ENSG00000204644	breast	1.59×10-8 (p-value)	GTEx Portal V6	134

ZFP57	?	ENSG00000204644	tibial artery	1.18×10-9 (p-value)	GTEx Portal V6	150
HLA-H	?	ENSG00000206341	tibial artery	5.16×10-7 (p-value)	GTEx Portal V6	97
HLA-U	?	ENSG00000228078	tibial artery	1.32×10-5 (p-value)	GTEx Portal V6	11
HLA-J	?	ENSG00000204622	tibial artery	6.73×10-6 (p-value)	GTEx Portal V6	14
ZSCAN26	?	ENSG00000197062	thyroid	5.59×10-8 (p-value)	GTEx Portal V6	127
OR2B8P	?	ENSG00000182477	thyroid	8.80×10-9 (p-value)	GTEx Portal V6	99
ZFP57	?	ENSG00000204644	thyroid	2.70×10-12 (p-value)	GTEx Portal V6	155
OR2B7P	?	ENSG00000187763	thyroid	9.85×10-8 (p-value)	GTEx Portal V6	96
ZNF192P1	?	ENSG00000226314	thyroid	4.88×10-6 (p-value)	GTEx Portal V6	68
RP1-265C24.5	?	ENSG00000219392	thyroid	1.41×10-5 (p-value)	GTEx Portal V6	1
RP1-265C24.5	?	ENSG00000219392	skeletal muscle	7.35×10-7 (p-value)	GTEx Portal V6	99
AL022393.7	?	ENSG00000216901	skeletal muscle	1.86×10-11 (p-value)	GTEx Portal V6	113
ZSCAN23	?	ENSG00000187987	skeletal muscle	5.54×10-6 (p-value)	GTEx Portal V6	69
ZFP57	?	ENSG00000204644	skeletal muscle	9.19×10-8 (p-value)	GTEx Portal V6	83
HLA-H	?	ENSG00000206341	skeletal muscle	2.11×10-6 (p-value)	GTEx Portal V6	89
HLA-J	?	ENSG00000204622	skeletal muscle	9.59×10-8 (p-value)	GTEx Portal V6	67
ZFP57	?	ENSG00000204644	liver	9.11×10-7 (p-value)	GTEx Portal V6	7
OR2B8P	?	ENSG00000182477	sun exposed skin	6.50×10-8 (p-value)	GTEx Portal V6	97
ZFP57	?	ENSG00000204644	sun exposed skin	4.62×10-10 (p-value)	GTEx Portal V6	154
HLA-H	?	ENSG00000206341	sun exposed skin	1.74×10-5 (p-value)	GTEx Portal V6	1
ZFP57	?	ENSG00000204644	unexposed skin	1.59×10-7 (p-value)	GTEx Portal V6	121
RP1-265C24.5	?	ENSG00000219392	left ventricle	5.52×10-10 (p-value)	GTEx Portal V6	104
AL022393.7	?	ENSG00000216901	left ventricle	1.04×10-5 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	left ventricle	1.56×10-6 (p-value)	GTEx Portal V6	17
ZNF165	?	ENSG00000197279	left ventricle	8.89×10-6 (p-value)	GTEx Portal V6	2
ZNF192P1	?	ENSG00000226314	left ventricle	6.32×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	aorta	2.84×10-9 (p-value)	GTEx Portal V6	154
ZFP57	?	ENSG00000204644	subcutaneous adipocytes	4.22×10-8 (p-value)	GTEx Portal V6	144
RP1-265C24.5	?	ENSG00000219392	subcutaneous adipocytes	3.75×10-6 (p-value)	GTEx Portal V6	55
HIST1H4J	?	ENSG00000197238	subcutaneous adipocytes	3.92×10-6 (p-value)	GTEx Portal V6	2
ZFP57	?	ENSG00000204644	tibial nerve	1.32×10-8 (p-value)	GTEx Portal V6	143
AL022393.7	?	ENSG00000216901	tibial nerve	3.35×10-6 (p-value)	GTEx Portal V6	60
ZSCAN26	?	ENSG00000197062	esophagus mucosa	7.64×10-6 (p-value)	GTEx Portal V6	37
ZFP57	?	ENSG00000204644	esophagus mucosa	1.19×10-14 (p-value)	GTEx Portal V6	155
ZNRD1	?	ENSG00000066379	esophagus mucosa	7.11×10-6 (p-value)	GTEx Portal V6	1
AL022393.7	?	ENSG00000216901	cerebellum	4.16×10-6 (p-value)	GTEx Portal V6	104
ZNF192P1	?	ENSG00000226314	cerebellum	2.00×10-5 (p-value)	GTEx Portal V6	1

ZFP57	?	ENSG00000204644	stomach	2.15×10-7 (p-value)	GTEx Portal V6	131
HIST1H3J	?	ENSG00000197153	testis	1.40×10-5 (p-value)	GTEx Portal V6	1
ZDHHC20P1	?	ENSG00000233916	testis	8.22×10-7 (p-value)	GTEx Portal V6	14
RP1-313I6.12	?	ENSG00000272009	testis	4.58×10-6 (p-value)	GTEx Portal V6	14
AL022393.7	?	ENSG00000216901	testis	1.77×10-5 (p-value)	GTEx Portal V6	10
XXbac-BPG308K3.5	?	ENSG00000225173	testis	3.21×10-5 (p-value)	GTEx Portal V6	1
ZSCAN26	ENST00000421553	ILMN_1760982	blood	2.25×10-6 (p-value)	MuTHER consortium	37
ZSCAN26	ENST00000614088					
ZSCAN26	ENST00000316606					
ZSCAN26	ENST00000619937					
ZSCAN31	ENST00000396838	ILMN_1655748	skin	4.43×10-8 (p-value)	MuTHER consortium	41
ZSCAN31	ENST00000344279		adipocyte	1.04×10-5 (p-value)	MuTHER consortium	41
ZSCAN31	ENST00000414429					
ZSCAN31	ENST00000439158					
ZSCAN31	ENST00000611469					
ZSCAN31	ENST00000446474					
HLA-V	ENST00000446817	ILMN_1660923	blood	1.84×10-7 (p-value)	MuTHER consortium	48
HLA-V	ENST00000429037		adipocyte	4.32×10-6 (p-value)	MuTHER consortium	48
?	?	ILMN_1654987	adipocyte	1.04×10-6 (p-value)	MuTHER consortium	39
			blood	7.02×10-6 (p-value)		
TRIM31	ENST00000376734	ILMN_1748685	skin	5.48×10-6 (p-value)	MuTHER consortium	11
TRIM31	ENST00000484583					
TRIM31	ENST00000471695					
TRIM31	ENST00000468264					
HLA-L	ENST00000463348	ILMN_1675282	blood	2.00×10-6 (p-value)	MuTHER consortium	21
ZNRD1	ENST00000471008	ILMN_2398587	blood	1.50×10-5 (p-value)	MuTHER consortium	2
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000463141					
?	?	ILMN_1679389	monocyte	7.45×10-16 (p-value)	Zeller et al.	10
?	?	ILMN_1666078	monocyte	9.45×10-29 (p-value)	Zeller et al.	10
BTN3A2	ENST00000396934	ILMN_1676528	monocyte	7.87×10-51 (p-value)	Zeller et al.	10
BTN3A2	ENST00000508906					
BTN3A2	ENST00000356386					
?	?	ILMN_1820787	monocyte	1.88×10-32 (p-value)	Zeller et al.	10
BTN3A2	ENST00000524459	ILMN_1700067	monocyte	1.85×10-37 (p-value)	Zeller et al.	10
BTN3A2	ENST00000528541					
BTN3A3	ENST00000467524					
BTN3A3	ENST00000482451					
BTN3A3	ENST00000494393					
BTN3A2	ENST00000527417					
BTN3A3	ENST00000244519					
BTN3A2	ENST00000528222					
BTN3A3	ENST00000474790					

BTN3A2	ENST00000377708					
BTN3A3	ENST00000496719					
BTN3A2	ENST00000396948					
BTN3A2	ENST00000527422					
BTN3A2	ENST00000356386					
?	?	ILMN_1883997	monocyte	3.70×10-33 (p-value)	Zeller et al.	10
?	?	ILMN_1671054	monocyte	6.26×10-38 (p-value)	Zeller et al.	9
HLA-G	ENST00000376828	ILMN_1656670	monocyte	2.84×10-16 (p-value)	Zeller et al.	9
HLA-G	ENST00000360323					
HLA-G	ENST00000478519					
HLA-G	ENST00000478355					
HLA-G	ENST00000428701					
HLA-G	ENST00000376815					
HLA-G	ENST00000376818					
ZFP57	?	ENSG00000204644	visceral adipocytes	2.25×10-6 (p-value)	GTEx Portal V6	27
ZFP57	?	ENSG00000204644	pancreas	3.20×10-6 (p-value)	GTEx Portal V6	17
ZSCAN26	?	ENSG00000197062	transverse colon	5.52×10-6 (p-value)	GTEx Portal V6	11
ZFP57	?	ENSG00000204644	transverse colon	5.30×10-7 (p-value)	GTEx Portal V6	14
ABT1	ENST00000274849	ILMN_1658083	monocyte	6.95×10-5 (p-value)	Fairfax et al.	2
SLC44A4	ENST00000487680	ILMN_1719250	b-cell	3.95×10-4 (p-value)	Fairfax et al.	1
SLC44A4	ENST00000544672					
SLC44A4	ENST00000229729					
SLC44A4	ENST00000375562					
ZNRD1	ENST00000471008	ILMN_2398587	b-cell	1.40×10-4 (p-value)	Fairfax et al.	1
ZNRD1	ENST00000359374					
ZNRD1-AS1_3	ENST00000611686					
ZNRD1	ENST00000376785					
ZNRD1	ENST00000376782					
ZNRD1	ENST00000332435					
ZNRD1	ENST00000463141					
C2	ENST00000486124	ILMN_1710740	monocyte	1.43×10-4 (p-value)	Fairfax et al.	1
C2	ENST00000469372					
CFB	ENST00000425368					
C2	ENST00000497706					
C2	ENST00000442278					
C2	ENST00000452323					
C2	ENST00000383177					
C2	ENST00000299367					
C2	ENST00000482060					
RP1-313I6.12	?	ENSG00000272009	adrenal gland	1.12×10-5 (p-value)	GTEx Portal V6	1
ZSCAN26	?	ENSG00000197062	adrenal gland	8.83×10-6 (p-value)	GTEx Portal V6	1
HLA-A	?	ENSG00000206503	brain	2.27×10-5 (q-value)	SeeQTL DB (Myers et al.)	6
GABBR1	ENST00000491829	ILMN_2395375	blood	3.40×10-5 (p-value)	Westra et al.	4
GABBR1	ENST00000377012					
GABBR1	ENST00000377034					
GABBR1	ENST00000478931					
GABBR1	ENST00000472823					
GABBR1	ENST00000377016					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
BTN3A2	?	ENSG00000186470	chr6	blood	3.72×10-6 (q-value)	SeeQTL DB (HapMap)	25
?	?	ILMN_1671054	chr6	monocyte	7.32×10-38 (p-value)	Zeller et al.	1
?	?	ILMN_1752592	chr6	monocyte	5.21×10-20 (p-value)	Zeller et al.	10
HLA-G	ENST00000376828	ILMN_1656670	chr6	monocyte	7.70×10-15 (p-value)	Zeller et al.	1
HLA-G	ENST00000360323						
HLA-G	ENST00000478519						
HLA-G	ENST00000478355						
HLA-G	ENST00000428701						
HLA-G	ENST00000376815						
HLA-G	ENST00000376818						
HLA-C	ENST00000470363	ILMN_2150787	chr6	monocyte	4.47×10-15 (p-value)	Zeller et al.	10
HLA-C	ENST00000383329						
HLA-C	ENST00000487245						
HLA-C	ENST00000376228						
HLA-C	ENST00000376237						
HLA-C	ENST00000620806						
HLA-C	ENST00000466892						
BTN3A2	ENST00000524682	209846_s_at	chr6	blood	1.50×10-12 (p-value)	Dixon et al.	6
BTN3A2	ENST00000604202						
BTN3A2	ENST00000396934						
BTN3A2	ENST00000532294						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000377708						
BTN3A2	ENST00000527422						
BTN3A2	ENST00000531055						
BTN3A2	ENST00000396948						
BTN3A2	ENST00000356386						
BTN3A2	ENST00000396934	212613_at	chr6	blood	8.90×10-10 (p-value)	Dixon et al.	6
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000244519	204820_s_at	chr6	blood	4.50×10-12 (p-value)	Dixon et al.	6
BTN3A2	ENST00000396934						
BTN3A2	ENST00000508906						
BTN3A2	ENST00000356386						
BTN3A3	ENST00000480110						
HLA-A	?	ENSG00000206503	chr6	brain	3.16×10-3 (q-value)	SeeQTL DB (Myers et al.)	2

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000443678	2	ENCP00000047794	OR2U1P
			OR2U1P
			OR2U1P
			OR2B4P
			OR2B4P
			OR2B4P
			OR2B4P
			OR2U1P

			OR2U1P OR2B4P OR2B4P OR2B4P OR2U1P
ENCE00000443629	1	ENCP00000047791	OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P OR2H4P
		ENCP00000047763	GPX5 GPX5
ENCE00000443616	1	ENCP00000047762	GPX6 GPX6
ENCE00000443718	1	ENCP00000047788	OR2P1P OR2P1P XXbac-BCX196D17.5 XXbac-BCX196D17.5 OR2B3 OR2B3 XXbac-BCX196D17.5 OR2B3 OR2P1P OR2B3 XXbac-BCX196D17.5 OR2B3 OR2P1P OR2P1P OR2B3 OR2B3 OR2P1P OR2B3 XXbac-BCX196D17.5 AL662791.1 OR2P1P XXbac-BCX196D17.5 XXbac-BCX196D17.5
ENCE00000443832	1	ENCP00000047793	OR2G1P OR2G1P OR2G1P OR2G1P OR2G1P OR2G1P
		ENCP00000047802	OR12D3 OR12D3 OR12D3 OR12D3 OR12D3 OR12D3 OR12D3
ENCE00000443781	1	ENCP00000047777	TRIM27 TRIM27 TRIM27 TRIM27 TRIM27 TRIM27 TRIM27
		ENCP00000047786	OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P OR2AD1P
		ENCP00000047778	TRIM27 TRIM27 TRIM27 TRIM27

				TRIM27
				TRIM27
				TRIM27
			ENCP00000047851	HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
				HLA-H
			ENCP00000047789	OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
				OR2N1P
			ENCP00000047824	GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
				GABBR1
ENCE00000443963		1	ENCP00000047854	DDX39BP1
				DDX39BP1
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				DDX39BP1
				DDX39BP1
				DDX39BP1
			ENCP00000047817	GABBR1
				GABBR1
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				GABBR1
				GABBR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494418 (promoter)	1	embryonic stem cell (H1ESC)	H3K9ac, CTCF, Yy1, HDAC2, TAF1, H3K4me2, H3K4me3, DNase1, Sin3Ak20, TAF7, H3K27me3, PolII, Rad21, Egr1
		HSMMtube	H3K4me3, H3K4me2, CTCF, H3K27me3, H2AZ, DNase1
		blood (K562)	H3K4me3, DNase1, CTCF, Ini1, PolIII, Egr1, H3K27ac, Max, Rad21, H3K9ac, Cmyc, H2AZ, PolII, H3K4me2
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, H3K27me3, DNase1
		liver (HepG2)	DNase1, H3K27me3, Rad21, H2AZ, H3K4me2, CTCF, Cmyc, H3K4me3
		blood (GM12878)	H3K4me3, CTCF, H2AZ, DNase1, Egr1, Yy1, Rad21
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	H3K4me2, H3K4me3, CTCF, DNase1
		NHLF	H3K4me3, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K4me2, CTCF, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K27ac, H3K4me3, PolII, Max, CTCF, DNase1, H3K9ac, H3K4me2
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF, Max, H3K4me2, H3K4me3
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000264210 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K27me3, CTCF, PolII, Rad21, H3K9ac, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	DNase1, H2AZ, H3K4me2
		blood (K562)	HEY1, H2AZ, Cfos, PolII, H3K4me2, CTCF, H3K4me3, Cmyc, H3K9ac, Ini1, Brg1, DNase1, PolIII, Egr1, H3K27ac, Max, ATF3, Rad21, E2F6

		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	H3K4me3, H3K4me2, DNase1, H2AZ
		liver (HepG2)	PolII, DNase1, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		blood (GM12878)	H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K4me2, H3K9ac, PolII
		nervous (NH-A)	H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	CTCF, DNase1, H3K4me2, H3K4me3
		NHLF	DNase1, H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H3K27ac, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, DNase1, H3K4me3
		cervix (HeLa-S3)	Max, H3K4me3, CTCF, DNase1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, PolII, CTCF, DNase1
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00000662255 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		blood (GM12878)	PolII, Cfos
		nervous (NH-A)	H3K27me3
		blood (DND-41)	H3K4me1
ENSR00000487496 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		HSMMtube	H3K27me3
		liver (HepG2)	H3K4me3, DNase1
		Osteobl	H3K27me3
		blood (DND-41)	H3K4me1
ENSR00000264215 (promoter)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3, H4K5ac, CTCF, PolII, TAF7, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3
		HSMMtube	H3K9ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K27ac
		blood (K562)	Ini1, Brg1, PolIII, H3K27ac, ZBTB33, Max, ATF3, H3K9ac, ETS1, HEY1, H2AZ, TAF1, Jund, Cjun, PolII, GTF2B, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, TAF1, ELF1, H2AZ, ZBTB33, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3, DNase1, Jund
		blood (GM12878)	PolII, H2AZ, ETS1, DNase1, CTCF, Yy1, ELF1, H3K4me3, ZBTB33, H3K27ac, H3K9ac
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me3, H3K27ac, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, CTCF
		cervix (HeLa-S3)	H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, PolII, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	Cmyc, DNase1, Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
ENSR00001702905 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1
		Osteobl	H3K4me2, H3K4me3
		blood (DND-41)	H3K4me2, H3K4me3
		blood (K562)	PolII, H3K4me3, H3K27ac, H3K9ac
		cervix (HeLa-S3)	H3K4me2, H3K4me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K4me3
		endothelium (HUVEC)	PolII
		liver (HepG2)	PolII
		A549	H3K36me3, H3K4me3
ENSR00001494427 (promoter flanking region)	1	endothelium (HUVEC)	H3K27me3
		blood (DND-41)	H3K27me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1
ENSR00000662257 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, PolII
		blood (DND-41)	H3K27me3
		blood (K562)	DNase1, HEY1, PolII
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1

		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		liver (HepG2)	DNase1, FOSL2, PolII, H3K4me1, FOXA1, H3K27ac
		nervous (NH-A)	DNase1
		A549	DNase1
		skin (NHEK)	DNase1
ENSR00000487504 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K4me3, DNase1, H3K4me2, Yy1, H3K27me3
		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2
		blood (K562)	DNase1, PolIII, H3K27ac, Max, H3K9ac, H2AZ, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me2, DNase1
		liver (HepG2)	H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K27me3
		blood (GM12878)	DNase1, H2AZ, Yy1, H3K4me3, H3K4me2
		lung (IMR90)	DNase1, H3K4me2, H3K4me3
		nervous (NH-A)	H3K4me3, H3K4me2
		skin (NHEK)	DNase1, H3K4me2
		NHLF	H3K4me3
		Osteobl	H3K4me2, H3K4me3, H2AZ
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K4me2
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K4me3, H3K27me3, DNase1, H3K4me2
		A549	DNase1, H3K4me2, H3K4me3
ENSR00001494434 (CTCF binding site)	1	NHLF	CTCF
		embryonic stem cell (H1ESC)	PolII, Rad21, CTCF, ATF3, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K27me3, DNase1
		Osteobl	H3K4me2
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K27me3
		endothelium (HUVEC)	CTCF
		blood (GM12878)	CTCF
		lung (IMR90)	H3K4me3, CTCF
		skin (NHEK)	CTCF, DNase1
ENSR00000662259 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K27me3
		embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF
		endothelium (HUVEC)	H3K27me3
		HSMMtube	H3K27me3
		lung (IMR90)	H3K27me3
		Osteobl	H3K27me3
		breast (HMEC)	H3K4me3, H3K4me2
ENSR00000487523 (enhancer)	1	embryonic stem cell (H1ESC)	DNase1
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL645937.1	upstream gene variant	1970	ENST00000610295	?	?	1
AL662791.1	downstream gene variant	2606	ENST00000625416	?	?	1
GABBR1	downstream gene variant	3852	ENST00000355973	?	ENSP00000348248	1
HCG14	upstream gene variant, downstream gene variant	1429	ENST00000444986	?	?	3
HCG15	downstream gene variant	1878	ENST00000438190	?	?	2
HCG16	upstream gene variant, downstream gene variant	284	ENST00000445544	?	?	3
KRT18P1	downstream gene variant, upstream gene variant	1046	ENST00000452408	?	?	2
LINC01556	upstream gene variant, downstream gene variant	4280	ENST00000377186	?	?	2
MAS1L	downstream gene variant, upstream gene variant	1255	ENST00000377127	NM_052967.1	ENSP00000366331	3
MAS1LP1	downstream gene variant	101	ENST00000419416	?	?	1
OR10C1	downstream gene variant	2678	ENST00000622521	?	ENSP00000481429	2

OR10C1	downstream gene variant	2701	ENST00000444197	?	ENSP00000419119	2
OR10C1	downstream gene variant	2701	ENST00000623086	NM_013941.3	ENSP00000485032	2
OR12D3	downstream gene variant	457	ENST00000396806	NM_030959.2	ENSP00000380023	1
OR14J1	upstream gene variant	331	ENST00000377160	NM_030946.1	ENSP00000366365	1
OR2AD1P	downstream gene variant, upstream gene variant	2298	ENST00000441992	?	?	2
OR2B3	upstream gene variant	2549	ENST00000377173	NM_001005226.2	ENSP00000366378	1
OR2B4P	upstream gene variant, downstream gene variant	1001	ENST00000356293	?	?	2
OR2H1	downstream gene variant	68	ENST00000377136	?	ENSP00000366340	3
OR2H1	downstream gene variant	3310	ENST00000484371	?	?	1
OR2H1	downstream gene variant	68	ENST00000377133	NM_030883.3	ENSP00000366337	3
OR2H1	downstream gene variant	1676	ENST00000473369	?	?	2
OR2H1	downstream gene variant	1676	ENST00000484554	?	?	2
OR2H1	downstream gene variant	1663	ENST00000396792	?	ENSP00000380010	2
OR2H1	downstream gene variant	68	ENST00000377132	?	ENSP00000366336	3
OR2H4P	upstream gene variant	126	ENST00000415595	?	?	2
OR2I1P	upstream gene variant, downstream gene variant	1442	ENST00000453522	?	?	2
OR2J2	downstream gene variant	4352	ENST00000377167	NM_030905.2	ENSP00000366372	1
OR2J3	upstream gene variant	4580	ENST00000377169	NM_001005216.3	ENSP00000366374	1
OR2J4P	upstream gene variant, downstream gene variant	893	ENST00000453647	?	?	3
OR2N1P	upstream gene variant	2604	ENST00000421799	?	?	1
OR2P1P	upstream gene variant	4325	ENST00000413771	?	?	1
OR2U2P	upstream gene variant	3180	ENST00000465951	?	?	1
OR2W1	downstream gene variant, upstream gene variant	553	ENST00000377175	NM_030903.3	ENSP00000366380	3
RN7SL471P	downstream gene variant	2542	ENST00000580476	?	?	1
RNU6-930P	upstream gene variant, downstream gene variant	126	ENST00000390938	?	?	3
RPL13P	upstream gene variant, downstream gene variant	1197	ENST00000396818	?	?	9
RPS17P1	downstream gene variant, upstream gene variant	788	ENST00000396783	?	?	3
SAR1P1	upstream gene variant	1473	ENST00000455287	?	?	1
TRIM27	downstream gene variant, upstream gene variant	140	ENST00000467742	?	?	3
TRIM27	downstream gene variant, upstream gene variant	1808	ENST00000377194	?	ENSP00000366399	3
TRIM27	downstream gene variant, upstream gene variant	1807	ENST00000377199	NM_006510.4	ENSP00000366404	3
TRIM27	downstream gene variant, upstream gene variant	1883	ENST00000481474	?	?	3
TRIM27	downstream gene variant, upstream gene variant	1837	ENST00000498117	?	?	3
TRIM27	downstream gene variant, upstream gene variant	481	ENST00000414543	?	ENSP00000400058	3
UBD	downstream gene variant	3738	ENST00000377050	NM_006398.3	ENSP00000366249	1
UBDP1	downstream gene variant	200	ENST00000457888	?	?	1
XXbac-BCX196D17.5	upstream gene variant, downstream gene	1976	ENST00000419702	?	?	2

	variant					
XXbac-BPG13B8.10	upstream gene variant	2243	ENST00000436804	?	?	1
XXbac-BPG13B8.11	upstream gene variant	2523	ENST00000618350	?	?	2
XXbac-BPG258E24.10	upstream gene variant	2485	ENST00000623946	?	?	1
XXbac-BPG258E24.10	upstream gene variant, downstream gene variant	400	ENST00000610326	?	?	4
XXbac-BPG258E24.10	upstream gene variant, downstream gene variant	445	ENST00000623334	?	?	3
XXbac-BPG308J9.3	upstream gene variant, downstream gene variant	1805	ENST00000606648	?	?	2
XXbac-BPG308J9.3	upstream gene variant	1778	ENST00000441381	?	?	1
XXbac-BPG308K3.5	downstream gene variant, upstream gene variant	958	ENST00000457253	?	?	8
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	46	ENST00000606624	?	?	6
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	157	ENST00000606727	?	?	9
XXbac-BPG308K3.6	downstream gene variant, upstream gene variant	484	ENST00000440244	?	?	8
ZNF311	downstream gene variant, upstream gene variant	1228	ENST00000377179	NM_001010877.2	ENSP00000366384	3
ZNF311	downstream gene variant, upstream gene variant	1172	ENST00000483450	?	?	3
ZNF90P2	downstream gene variant, upstream gene variant	804	ENST00000419323	?	?	6

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
UBD	ENST00000377050	NM_006398.3	ENSP00000366249	Y	taC/taT	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
HCG14	ENST00000444986	?	?	1
HCG15	ENST00000438190	?	?	1
OR11A1	ENST00000377149	?	ENSP00000366354	3
OR5V1	ENST00000377154	?	ENSP00000366359	4
TRIM27	ENST00000414543	?	ENSP00000400058	1
TRIM27	ENST00000377199	NM_006510.4	ENSP00000366404	5
TRIM27	ENST00000481474	?	?	5
TRIM27	ENST00000377194	?	ENSP00000366399	5
TRIM27	ENST00000496091	?	?	3
TRIM27	ENST00000498117	?	?	3
UBDP1	ENST00000457888	?	?	2
XXbac-BCX196D17.5	ENST00000419702	?	?	1
XXbac-BPG13B8.10	ENST00000436804	?	?	1
XXbac-BPG258E24.10	ENST00000623334	?	?	6
XXbac-BPG258E24.10	ENST00000623946	?	?	6
XXbac-BPG308J9.3	ENST00000606648	?	?	10
XXbac-BPG308J9.3	ENST00000441381	?	?	6

XXbac-BPG308K3.5	ENST00000457253	?	?	1
XXbac-BPG308K3.6	ENST00000440244	?	?	1
XXbac-BPG308K3.6	ENST00000606624	?	?	1
XXbac-BPG308K3.6	ENST00000606727	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GABBR1	ENST00000355973	?	ENSP00000348248	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
RPL13P	ENST00000396818	?	1
SAR1P1	ENST00000455287	?	1
TRIM27	ENST00000481474	?	1
XXbac-BPG308K3.6	ENST00000440244	?	2
XXbac-BPG308K3.6	ENST00000606727	?	1
ZNF90P2	ENST00000419323	?	4