Locus 317 (rs7083)

SNiPAcard

Block annotations

Block info

genomic range	chr11:117,106,280-117,163,765
block size	57,486 bp
variant count	40 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.709 [-3.083 – 0.486]	**gene(s) hit or close-by**	BACE1, BACE1-AS, CMB9-94B1.2, PCSK7, RNF214, SCARNA11
phastCons	μ = 0.072 [0 – 0.446]	**eQTL gene(s)**	BACE1, PCSK7, RNF214, RP11-109L13.1, SIDT2, TAGLN
GERP++	μ = -0.418 [-9.68 – 1.67]	**potentially regulated gene(s)**	–
CADD score	μ = 4.405 [0.738 – 12.24]	**disease gene(s)**	–

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-109L13.1	?	ENSG00000254851	sigmoid colon	1.19×10^{-6} (p-value)	GTEx Portal V6	25
RNF214	?	ENSG00000167257	subcutaneous adipocytes	4.10×10^{-6} (p-value)	GTEx Portal V6	9
RP11-109L13.1	?	ENSG00000254851	subcutaneous adipocytes	6.06×10^{-6} (p-value)	GTEx Portal V6	3
TAGLN	ENST00000525531	ILMN_1778668	blood	1.05×10^{-7} (p-value)	MuTHER consortium	10
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					
TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	blood	8.90×10^{-6} (p-value)	Westra et al.	10
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
PCSK7	ENST00000540028	ILMN_1699545	blood	5.70×10^{-7} (p-value)	Westra et al.	5
PCSK7	ENST00000529458					
PCSK7	ENST00000534529					
PCSK7	ENST00000320934					
PCSK7	ENST00000527037					
SIDT2	ENST00000278951	ILMN_1791912	monocyte	5.17×10^{-19} (p-value)	Zeller et al.	2
SIDT2	ENST00000532062					
SIDT2	ENST00000324225					
SIDT2	ENST00000431081					
SIDT2	ENST00000620360					
TAGLN	ENST00000525531	ILMN_1778668	monocyte	1.08×10^{-23} (p-value)	Zeller et al.	3
TAGLN	ENST00000532870					
TAGLN	ENST00000278968					
TAGLN	ENST00000392951					

TAGLN	ENST00000530649					
TAGLN	ENST00000533863					
TAGLN	ENST00000525531	ILMN_2400935	monocyte	6.54×10-17 (p-value)	Zeller et al.	2
TAGLN	ENST00000529622					
TAGLN	ENST00000278968					
TAGLN	ENST00000530649					
TAGLN	ENST00000529792					
TAGLN	ENST00000532870					
TAGLN	ENST00000392951					
TAGLN	ENST00000533863					
BACE1	?	ENSG00000186318	testis	1.77×10-7 (p-value)	GTEx Portal V6	10
BACE1	ENST00000528053	ILMN_1797804	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
BACE1	ENST00000392937					
BACE1	ENST00000313005					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000321759 (open chromatin region)	1	endothelium (HUVEC)	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (K562)	DNase1
		A549	H3K36me3
ENSR00000569588 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		lung (IMR90)	H3K36me3
ENSR00001573947 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1
		HSMMtube	DNase1
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K27ac, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	DNase1
		A549	H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	H3K36me3
ENSR00000569591 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K36me3
		NHLF	H3K36me3, DNase1
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun, DNase1
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)

gene	variant type	imm(distance)	transcript	RefSeq id	protein	variant(s)
BACE1	downstream gene variant	1515	ENST00000528053	?	ENSP00000431848	1
BACE1	downstream gene variant	3651	ENST00000510630	NM_001207049.1	ENSP00000422461	1
BACE1	downstream gene variant	3699	ENST00000513780	NM_138972.3	ENSP00000424536	1
BACE1	downstream gene variant	3882	ENST00000392937	NM_001207048.1	ENSP00000475405	1
BACE1	downstream gene variant	822	ENST00000514464	?	?	1
BACE1	downstream gene variant, upstream gene variant	1052	ENST00000509916	?	?	2
BACE1	downstream gene variant	3699	ENST00000445823	NM_138971.3	ENSP00000403685	1
BACE1	downstream gene variant	22	ENST00000504995	?	ENSP00000434486	1
BACE1	downstream gene variant	3699	ENST00000428381	NM_138973.3	ENSP00000402228	1
BACE1	downstream gene variant	4798	ENST00000530824	?	?	1
BACE1	downstream gene variant	3383	ENST00000313005	NM_012104.4	ENSP00000318585	1
CMB9-94B1.2	downstream gene variant	3956	ENST00000613535	?	?	1
PCSK7	upstream gene variant	3039	ENST00000320934	NM_004716.2	ENSP00000325917	3
PCSK7	upstream gene variant	3469	ENST00000540028	?	ENSP00000441944	3
PCSK7	upstream gene variant	3039	ENST00000530269	?	ENSP00000433252	3
PCSK7	upstream gene variant	3696	ENST00000524507	?	ENSP00000433841	2
PCSK7	upstream gene variant	3490	ENST00000532301	?	ENSP00000436459	3
RNF214	downstream gene variant	148	ENST00000531452	NM_001077239.1	ENSP00000431643	1
RNF214	upstream gene variant, downstream gene variant	590	ENST00000534709	?	?	2
RNF214	downstream gene variant	150	ENST00000300650	NM_207343.3	ENSP00000300650	1
RNF214	downstream gene variant	672	ENST00000531287	NM_001278249.1	ENSP00000435361	1
RNF214	upstream gene variant, downstream gene variant	2632	ENST00000524917	?	?	2
RNF214	downstream gene variant	1996	ENST00000529869	?	?	2
SCARNA11	upstream gene variant, downstream gene variant	93	ENST00000517183	?	?	18

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1	ENST00000313005	NM_012104.4	ENSP00000318585	1
BACE1	ENST00000392937	NM_001207048.1	ENSP00000475405	1
BACE1	ENST00000513780	NM_138972.3	ENSP00000424536	1
BACE1	ENST00000428381	NM_138973.3	ENSP00000402228	1
BACE1	ENST00000510915	?	ENSP00000475171	1
BACE1	ENST00000445823	NM_138971.3	ENSP00000403685	1
BACE1	ENST00000530824	?	?	1
BACE1	ENST00000510630	NM_001207049.1	ENSP00000422461	1
BACE1	ENST00000528053	?	ENSP00000431848	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1-AS	ENST00000614401	?	?	1

RNF214	ENST00000524917	?	?	1
RNF214	ENST00000529869	?	?	6
RNF214	ENST00000530849	?	ENSP00000432903	38
RNF214	ENST00000531287	NM_001278249.1	ENSP00000435361	38
RNF214	ENST00000300650	NM_207343.3	ENSP00000300650	38
RNF214	ENST00000534428	?	ENSP00000434186	5
RNF214	ENST00000531452	NM_001077239.1	ENSP00000431643	38

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
BACE1	ENST00000313005	NM_012104.4	ENSP00000318585	1
BACE1	ENST00000392937	NM_001207048.1	ENSP00000475405	1
RNF214	ENST00000530849	?	ENSP00000432903	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
BACE1-AS	ENST00000618857	?	1
RNF214	ENST00000534709	?	1