Locus 42 (rs4725617)

SNiPAcard

Block annotations

Block info

genomic range	chr7:143,092,918-143,108,081
block size	15,164 bp
variant count	10 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.574 [-1.838 – 0.994]	gene(s) hit or close-by	EPHA1, EPHA1-AS1, ZYX
phastCons	μ = 0.026 [0 – 0.13]	eQTL gene(s)	EPHA1, EPHA1-AS1, ZYX
GERP++	μ = 0.477 [-1.57 – 3.88]	potentially regulated gene(s)	–
CADD score	μ = 5.976 [2.371 – 12.8]	disease gene(s)	–

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)
EPHA1	missense variant	ENST00000275815	NM_005232.4	ENSP00000275815	V/A	gTg/gCg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
EPHA1-AS1	?	ENSG00000229153	blood	3.02×10^{-8} (p-value)	GTEx Portal V6	2
ZYX	?	ENSG00000159840	blood	2.45×10^{-8} (p-value)	GTEx Portal V6	3
EPHA1	?	ENSG00000146904	sun exposed skin	2.87×10^{-7} (p-value)	GTEx Portal V6	8
EPHA1	?	ENSG00000146904	thyroid	6.98×10^{-7} (p-value)	GTEx Portal V6	8

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001564739 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	DNase1, CTCF
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1
		endothelium (HUVEC)	DNase1, H3K4me1, PolII
		liver (HepG2)	H3K4me1, H3K4me2, HNF4A, DNase1, RXRA
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000069717 (CTCF binding site)	2	embryonic stem cell (H1ESC)	CTCF, Rad21, H3K36me3
		blood (K562)	CTCF, H3K27me3
		skin (NHDF-AD)	CTCF
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	DNase1, PolII, H3K4me1

		liver (HepG2)	RXRA, H3K4me1, H3K4me2, HNF4A, DNase1
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000635676 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K27me3
		A549	H3K27me3
		blood (K562)	H3K27me3
		skin (NHEK)	H3K36me3
ENSR00001718722 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1, H3K4me3, PolII, Rad21, Egr1, Nrsf, CTCF, TAF1, SP1
		HSMMtube	H3K27me3
		blood (K562)	H3K27me3, Egr1, CTCF, DNase1
		breast (HMEC)	CTCF, H3K4me3
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac, H3K4me3
		endothelium (HUVEC)	CTCF
		liver (HepG2)	H3K4me3, H3K4me2, DNase1
		blood (GM12878)	ZEB1
		lung (IMR90)	CTCF
		nervous (NH-A)	H3K27me3
		A549	H3K4me3, H3K4me2, H3K27me3
		skin (NHEK)	DNase1, CTCF, H3K4me3
ENSR00000635678 (promoter flanking region)	2	monocytes (Monocytes-CD14+)	H3K4me3, H3K27ac, H3K4me1, DNase1
		embryonic stem cell (H1ESC)	H3K4me2, DNase1
		Osteobl	H3K4me2
		blood (K562)	H3K27me3
		skin (NHDF-AD)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
EPHA1	upstream gene variant	4434	ENST00000458129	?	?	2
EPHA1	downstream gene variant, upstream gene variant	819	ENST00000497891	?	?	6
EPHA1	downstream gene variant, upstream gene variant	1267	ENST00000479459	?	?	4
EPHA1	upstream gene variant	224	ENST00000465208	?	?	3
EPHA1	upstream gene variant	1041	ENST00000275815	NM_005232.4	ENSP00000275815	4
EPHA1	upstream gene variant	1128	ENST00000488068	?	?	4
EPHA1	upstream gene variant	3326	ENST00000494989	?	?	2
EPHA1-AS1	upstream gene variant	1526	ENST00000429289	?	?	2
EPHA1-AS1	upstream gene variant	1546	ENST00000421648	?	?	2
ZYX	downstream gene variant	4716	ENST00000354434	?	ENSP00000346417	1
ZYX	downstream gene variant	4714	ENST00000322764	NM_003461.4, NM_001010972.1	ENSP00000324422	1
ZYX	downstream gene variant	4716	ENST00000392910	?	ENSP00000376642	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
EPHA1	ENST00000497891	?	?	3
EPHA1	ENST00000488068	?	?	5
EPHA1	ENST00000275815	NM_005232.4	ENSP00000275815	5
EPHA1	ENST00000494989	?	?	2

EPHA1-AS1	ENST00000421648	?	?	4
EPHA1-AS1	ENST00000429289	?	?	4

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
EPHA1	ENST00000497891	?	1
EPHA1	ENST00000488068	?	2