Locus 77 (rs1250258)

SNiPAcard

Block annotations

Block info

genomic range	chr2:216,287,093-216,306,635
block size	19,543 bp
variant count	15 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.022 [-1.29 – 3.387]	gene(s) hit or close-by	AC012462.1, FN1
phastCons	μ = 0.133 [0 – 1]	eQTL gene(s)	FN1
GERP++	μ = -0.320 [-7.88 – 5.35]	potentially regulated gene(s)	AC093850.2, FN1
CADD score	μ = 5.604 [0.051 – 22.4]	disease gene(s)	FN1

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
LDL cholesterol	<3.00×10-8	GWAS Catalog	24097068	1

Disease gene annotation

gene	trait	source DB	source entry/link
FN1	GLOMERULOPATHY WITH FIBRONECTIN DEPOSITS 2	OMIM	MIM:601894
FN1	Fibronectin glomerulopathy	OrphaNet	OrphaNet:84090

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)
FN1	missense variant	ENST00000359671	?	ENSP00000352696	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000432072	?	ENSP00000399538	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000357867	NM_212474.1	ENSP00000350534	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000421182	?	ENSP00000394423	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000446046	NM_212478.1	ENSP00000410422	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000443816	?	ENSP00000415018	Q/L	cAg/cTg	tolerated	unknown	1
FN1	missense variant	ENST00000336916	NM_002026.2	ENSP00000338200	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000354785	NM_212482.1	ENSP00000346839	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000323926	?	ENSP00000323534	Q/L	cAg/cTg	tolerated	benign	1
FN1	missense variant	ENST00000426059	NM_054034.2	ENSP00000398907	Q/L	cAg/cTg	?	benign	1
FN1	missense variant	ENST00000356005	NM_212476.1	ENSP00000348285	Q/L	cAg/cTg	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FN1	ENST00000426059	ILMN_1675646	b-cell	1.72×10^{-5} (p-value)	Fairfax et al.	1
?	?	ILMN_1673991	monocyte	5.73×10^{-13} (p-value)	Zeller et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000529793	1	ENST00000357867, ENST00000421182, ENST00000432072, ENST00000443816, ENST00000354785, ENST00000426059, ENST00000336916, ENST00000359671, ENST00000323926, ENST00000446046, ENST00000356005	FN1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000277616	2	ENCP00000029547	FN1
		ENCP00000029544	FN1
		ENCP00000029545	FN1
		ENCP00000029538	FN1
		ENCP00000029543	FN1
		ENCP00000029540	FN1
		ENCP00000029542	FN1
		ENCP00000029541	FN1
ENCE00000277620	1	ENCP00000029547	FN1
		ENCP00000029544	FN1
		ENCP00000029538	FN1
		ENCP00000029545	FN1
		ENCP00000029543	FN1
		ENCP00000029540	FN1
		ENCP00000029542	FN1
		ENCP00000029558	AC093850.2
		ENCP00000029541	FN1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001551486 (promoter flanking region)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (K562)	DNase1
		skin (NHDF-AD)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	Max, PolII, H3K36me3
		liver (HepG2)	PolII, H3K4me1, H3K4me2, H3K27ac, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3, H3K4me2, H4K20me1, H3K9ac
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000607008 (promoter)	4	embryonic stem cell (H1ESC)	ATF3, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1, Rad21, PolII, H3K36me3, H3K27me3
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K36me3
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me3, H3K27ac, H3K4me2

		liver (HepG2)	TAF1, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, H3K27me3, H3K36me3, DNase1, H3K79me2, PolII, Jund
		blood (GM12878)	DNase1, H2AZ, H3K4me3, H3K4me2
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K4ac, H3K9ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H4K20me1, H3K9ac
		skin (NHEK)	H3K27me3, H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K4me3, H3K4me2, H3K36me3, H3K27ac
		blood (DND-41)	H3K27me3, H3K4me3, H3K4me1
		breast (HMEC)	H3K36me3, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		monocytes (Monocytes-CD14+)	H3K27me3, H3K4me3
		endothelium (HUVEC)	H3K27ac, PolII, DNase1, H3K36me3, H3K9ac, H3K4me3, H3K4me2, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K36me3, H3K27me3
ENSR00001661218 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1
		skin (NHDF-AD)	DNase1
ENSR00001551491 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	Rad21
		blood (K562)	H2AZ
		skin (NHDF-AD)	DNase1
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		liver (HepG2)	H3K4me1, H2AZ, H3K4me2
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC012462.1	upstream gene variant, downstream gene variant	494	ENST00000412951	?	?	9
FN1	upstream gene variant	2257	ENST00000426059	NM_054034.2	ENSP00000398907	2
FN1	upstream gene variant	2262	ENST00000446046	NM_212478.1	ENSP00000410422	2
FN1	upstream gene variant	2262	ENST00000357867	NM_212474.1	ENSP00000350534	2
FN1	upstream gene variant	2262	ENST00000359671	?	ENSP00000352696	2
FN1	upstream gene variant	2263	ENST00000432072	?	ENSP00000399538	2
FN1	upstream gene variant	1100	ENST00000496542	?	?	2
FN1	upstream gene variant	2263	ENST00000421182	?	ENSP00000394423	2
FN1	upstream gene variant	2262	ENST00000336916	NM_002026.2	ENSP00000338200	2
FN1	upstream gene variant	2257	ENST00000443816	?	ENSP00000415018	2
FN1	upstream gene variant	2262	ENST00000323926	?	ENSP00000323534	2
FN1	upstream gene variant	2158	ENST00000354785	NM_212482.1	ENSP00000346839	3
FN1	upstream gene variant	2262	ENST00000356005	NM_212476.1	ENSP00000348285	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FN1	ENST00000359671	?	ENSP00000352696	10
FN1	ENST00000432072	?	ENSP00000399538	10
FN1	ENST00000357867	NM_212474.1	ENSP00000350534	10
FN1	ENST00000421182	?	ENSP00000394423	10

FN1	ENST00000323926	?	ENSP00000323534	10
FN1	ENST00000443816	?	ENSP00000415018	10
FN1	ENST00000426059	NM_054034.2	ENSP00000398907	10
FN1	ENST00000354785	NM_212482.1	ENSP00000346839	10
FN1	ENST00000446046	NM_212478.1	ENSP00000410422	10
FN1	ENST00000336916	NM_002026.2	ENSP00000338200	10
FN1	ENST00000356005	NM_212476.1	ENSP00000348285	10