Locus 272 (rs7730934)

SNiPAcard

Block annotations

Block info

genomic range	chr5:55,228,754-55,318,612
block size	89,859 bp
variant count	55 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.273 [-4.321 – 4.663]	gene(s) hit or close-by	AC008914.1, CTD-2031P19.4, CTD-2031P19.5, FLJ31104, IL6ST, snoU13
phastCons	μ = 0.053 [0 – 1]	eQTL gene(s)	IL6ST
GERP++	μ = -0.092 [-4.49 – 5.76]	potentially regulated gene(s)	ANKRD55, CTC-236F12.4, DDX4, IL6ST, RP11-365H8.2, SLC38A9
CADD score	μ = 4.278 [0.005 – 23.9]	disease gene(s)	ANKRD55

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ANKRD55	Oligoarticular juvenile arthritis	OrphaNet	OrphaNet:85410
ANKRD55	Juvenile rheumatoid factor-negative polyarthritis	OrphaNet	OrphaNet:85408

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)
IL6ST	missense variant	ENST00000381298	NM_002184.3	ENSP00000370698	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000381294	NM_001190981.1	ENSP00000370694	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000522633	?	ENSP00000435399	G/R	Ggt/Cgt	deleterious	benign	1
IL6ST	missense variant	ENST00000502326	?	ENSP00000462158	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000503773	?	ENSP00000426224	G/R	Ggt/Cgt	deleterious	benign	1
IL6ST	missense variant	ENST00000336909	?	ENSP00000338799	2	2	?, deleterious	?, benign	2
IL6ST	missense variant	ENST00000381287	NM_175767.2	ENSP00000370687	G/R	Ggt/Cgt	deleterious	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
IL6ST	?	ENSG00000134352	tibial nerve	3.31×10-8 (p-value)	GTEx Portal V6	20

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000697123	1	ENST00000381287, ENST00000381298, ENST00000381294, ENST00000336909	IL6ST

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000409418	1	ENCP00000044145	ANKRD55
ENCE00000409253	2	ENCP00000044146	RP11-365H8.2
		ENCP00000044129	DDX4
		ENCP00000044127	SLC38A9
		ENCP00000044151	CTC-236F12.4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001280383 (promoter flanking region)	1	NHLF	H3K36me3, DNase1
		embryonic stem cell (H1ESC)	CTCF, Rad21, DNase1
		HSMMtube	DNase1, CTCF
		Osteobl	H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1, CTCF
		breast (HMEC)	CTCF, DNase1
		muscle (HSMM)	CTCF, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H4K20me1, H3K36me3
		endothelium (HUVEC)	CTCF, H3K36me3
		liver (HepG2)	H3K79me2, Rad21, CTCF, H3K36me3, DNase1
		blood (GM12878)	Yy1, Cjun, BATF, Tcf12, CTCF, IRF4, Rad21, H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3, CTCF
		A549	H3K36me3
ENSR00001409454 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		Osteobl	H3K36me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		liver (HepG2)	H3K36me3, CTCF, H3K79me2, Rad21
		blood (GM12878)	CTCF, H3K79me2, H3K4me3
		lung (IMR90)	H3K36me3
		A549	H3K4me3, CTCF
		skin (NHEK)	CTCF
ENSR00001409459 (promoter)	4	embryonic stem cell (H1ESC)	H3K9ac, TAF1, H3K4me2, SP1, H3K4me3, Sin3Ak20, PolII, H3K27ac, CTCF, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		blood (K562)	TAF1, Cfos, PolII, H3K4me2, DNase1, H3K4me3, H2AZ, HEY1, H3K79me2, Egr1, H3K27ac, Max, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	PolII, H3K79me2, TAF1, H3K4me2, FOXA1, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac, H3K56ac
		blood (GM12878)	H3K9ac, H3K4me2, EBF1, Pbx3, PolII, H2AZ, Tcf12, DNase1, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, Cfos
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2, H3K4me3, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, CTCF
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H3K36me3
		blood (DND-41)	H3K4me2, H3K9ac, H3K4me3, H3K27me3
		breast (HMEC)	DNase1, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		cervix (HeLa-S3)	DNase1, H3K9ac, Jund, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc, Cjun
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001693442	1	blood (DND-41)	H3K27me3

(enhancer)		skin (NHDF-AD)	H3K4me1, DNase1, H3K27ac
ENSR00001280396 (enhancer)	3	blood (GM12878)	PU1
		blood (DND-41)	H3K27me3
		muscle (HSMM)	H3K36me3
ENSR00001409462 (promoter flanking region)	1	NHLF	DNase1, H3K27ac
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me2, H3K27ac, H3K4me1, DNase1, H3K9ac
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac, H3K4me2
		monocytes (Monocytes-CD14+)	H3K4me1
		liver (HepG2)	H3K4me1, HNF4A, H3K27ac
		lung (IMR90)	DNase1, H3K4me2
		nervous (NH-A)	DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
IL6ST	1	ENST00000336909 ENST00000381286 ENST00000381287 ENST00000381293 ENST00000381294 ENST00000381298 ENST00000502326 ENST00000503773 ENST00000506241 ENST00000522633	hsa-miR-296-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008914.1	upstream gene variant	4185	ENST00000624860	?	ENSP00000485310	1
CTD-2031P19.4	upstream gene variant, downstream gene variant	4110	ENST00000582508	?	?	2
CTD-2031P19.5	upstream gene variant	3217	ENST00000576302	?	?	1
FLJ31104	upstream gene variant, downstream gene variant	549	ENST00000500093	?	?	10
IL6ST	downstream gene variant, upstream gene variant	1285	ENST00000381287	NM_175767.2	ENSP00000370687	7
IL6ST	upstream gene variant	1609	ENST00000423954	?	?	4
IL6ST	downstream gene variant, upstream gene variant	2169	ENST00000381298	NM_002184.3	ENSP00000370698	3
IL6ST	downstream gene variant, upstream gene variant	1321	ENST00000396816	?	ENSP00000463896	10
IL6ST	downstream gene variant, upstream gene variant	1219	ENST00000381286	?	ENSP00000370686	8
IL6ST	downstream gene variant, upstream gene variant	1285	ENST00000336909	?	ENSP00000338799	7
IL6ST	downstream gene variant, upstream gene variant	420	ENST00000503773	?	ENSP00000426224	4
IL6ST	downstream gene variant, upstream gene variant	482	ENST00000502326	?	ENSP00000462158	4
IL6ST	downstream gene variant, upstream gene variant	1148	ENST00000583149	?	?	6
IL6ST	downstream gene variant, upstream gene variant	585	ENST00000506241	?	?	6
IL6ST	downstream gene variant, upstream gene variant	977	ENST00000523039	?	?	3
IL6ST	downstream gene variant, upstream gene variant	1816	ENST00000577363	?	?	6
IL6ST	downstream gene variant, upstream gene variant	1219	ENST00000381293	?	ENSP00000370693	8
IL6ST	upstream gene variant	339	ENST00000522633	?	ENSP00000435399	3
IL6ST	downstream gene variant, upstream gene variant	536	ENST00000381294	NM_001190981.1	ENSP00000370694	8
snoU13	downstream gene variant, upstream gene variant	89	ENST00000459441	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTD-2031P19.5	ENST00000576302	?	?	2
FLJ31104	ENST00000500093	?	?	3
IL6ST	ENST00000381294	NM_001190981.1	ENSP00000370694	22
IL6ST	ENST00000336909	?	ENSP00000338799	24
IL6ST	ENST00000577363	?	?	25
IL6ST	ENST00000583149	?	?	1
IL6ST	ENST00000381286	?	ENSP00000370686	24
IL6ST	ENST00000522633	?	ENSP00000435399	29
IL6ST	ENST00000381298	NM_002184.3	ENSP00000370698	37
IL6ST	ENST00000381293	?	ENSP00000370693	24
IL6ST	ENST00000502326	?	ENSP00000462158	35
IL6ST	ENST00000381287	NM_175767.2	ENSP00000370687	25
IL6ST	ENST00000506241	?	?	5
IL6ST	ENST00000396816	?	ENSP00000463896	12
IL6ST	ENST00000503773	?	ENSP00000426224	36

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IL6ST	ENST00000336909	?	ENSP00000338799	2
IL6ST	ENST00000522633	?	ENSP00000435399	1
IL6ST	ENST00000381298	NM_002184.3	ENSP00000370698	2
IL6ST	ENST00000381287	NM_175767.2	ENSP00000370687	3
IL6ST	ENST00000503773	?	ENSP00000426224	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
IL6ST	ENST00000506241	?	1
IL6ST	ENST00000423954	?	1