Locus 48 (rs103294)

SNiPAcard

Block annotations

Block info

genomic range	chr19:54,792,761-54,809,343
block size	16,583 bp
variant count	25 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.377 [-4.251 – 0.44]	**gene(s) hit or close-by**	AC008984.5, AC008984.7, AC010518.2, AC010518.3
phastCons	μ = 0.006 [0 – 0.089]	**eQTL gene(s)**	AC010518.2, ITIH6, LILRA1, LILRB2, TSEN34
GERP++	μ = -0.450 [-1.87 – 0.713]	**potentially regulated gene(s)**	AC008753.6, AC008984.5, AC008984.5, AC008984.7, AC008984.7, AC009892.10, AC009892.10, AC009892.5, AC010518.3, AC010518.3, AC011515.2, AC011515.2, AC012314.19, AC012314.19, AC012314.2, KIR2DL1, KIR2DL3, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR3DL3, LAIR1, LAIR1, LAIR1, LAIR1, LAIR1, LENG8, LENG8, LENG8, LENG8, LILRA1, LILRA1, LILRA1, LILRA1, LILRA2, LILRA2, LILRA2, LILRA2, LILRA3, LILRA3, LILRA3, LILRA3, LILRA5, LILRA5, LILRA5, LILRA5, LILRA5, LILRA6, LILRA6, LILRA6, LILRA6, LILRA6, LILRB1, LILRB1, LILRB1, LILRB1, LILRB1, LILRB2, LILRB2, LILRB2, LILRB2, LILRB2, LILRB3, LILRB3, LILRB3, LILRB3, LILRB4, LILRB4, LILRB4, LILRB4, LILRB4, MYADM, NLRP12, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, OSCAR, VN1R105P
CADD score	μ = 1.285 [0.008 – 5.356]	**disease gene(s)**	TSEN34, NLRP12

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
HDL cholesterol	$<3.00\times10^{-23}$	GWAS Catalog	24097068	1
PROSTATE CANCER	$<5.00\times10^{-16}$	GWAS Catalog	23023329	1
HDL cholesterol	$<4.00\times10^{-16}$	GWAS Catalog	20686565	1

Disease gene annotation

gene	trait	source DB	source entry/link
TSEN34	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
NLRP12	FAMILIAL COLD AUTOINFLAMMATORY SYNDROME 2	OMIM	MIM:611762
TSEN34	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
TSEN34	Pontocerebellar hypoplasia type 2	OrphaNet	OrphaNet:2524
NLRP12	NLRP12-associated hereditary periodic fever syndrome	OrphaNet	OrphaNet:247868

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRB2	ENST00000434421	ILMN_2312340	blood	1.93×10^{-5} (p-value)	Westra et al.	3
LILRB2	ENST00000493242					
LILRB2	ENST00000455108					
LILRB2	ENST00000314446					
LILRB2	ENST00000391748					
TSEN34	ENST00000396383	ILMN_1673111	blood	4.50×10^{-5} (p-value)	Westra et al.	2
TSEN34	ENST00000396388					
TSEN34	ENST00000302937					
TSEN34	ENST00000496583					
TSEN34	ENST00000396383	ILMN_2368292	blood	1.27×10^{-5} (p-value)	Westra et al.	4
TSEN34	ENST00000396388					
TSEN34	ENST00000302937					
TSEN34	ENST00000496583					
LILRB2	?	ENSG00000131042	sun exposed skin	1.90×10^{-8} (p-value)	GTEx Portal V6	22
LILRA1	ENST00000618665	ILMN_1661631	monocyte	2.83×10^{-289} (p-value)	Zeller et al.	2
LILRA1	ENST00000477255					
LILRA1	ENST00000495417					
AC010518.2	ENST00000615364					
?	?	ILMN_1786303	monocyte	4.40×10^{-265} (p-value)	Zeller et al.	2
LILRA1	ENST00000618665	ILMN_1661631	liver	4.88×10^{-15} (p-value)	Innocenti et al.	1
LILRA1	ENST00000477255					
AC010518.2	ENST00000615364					
LILRA1	ENST00000495417					

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ITIH6	ENST00000218436	ILMN_1709177	chrX	monocyte	6.95×10^{-24} (p-value)	Zeller 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)
ENCE00000233622	2	ENCP00000024752	LILRB3 LILRB3 LILRB3 LILRB3
ENCE00000233400	5	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024761	AC008984.5 AC008984.5
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024769	LAIR1 LAIR1

Locus 48 (sentinel SNP rs103294 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

	LAIR1 LAIR1 LAIR1
ENCP00000024758	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
ENCP00000024753	LILRA6 LILRA6 LILRA6 LILRA6 LILRA6
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024705	MYADM
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024721	AC012314.2
ENCP00000024722	OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR OSCAR
ENCP00000024703	NLRP12
ENCP00000024788	LENG8 LENG8 LENG8 LENG8
ENCP00000024803	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCP00000024808	VN1R105P
ENCP00000024773	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
ENCP00000024763	LILRA5 AC008984.7 AC008984.7 LILRA5 LILRA5 LILRA5

	LILRA5
ENCP00000024772	LAIR1
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ENCP00000024810	LILRB4
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ENCP00000024759	AC010518.3
	AC010518.3
ENCP00000024771	LAIR1
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ENCP00000024819	KIR2DL1
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ENCP00000024799	LILRA1
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	LILRA1
ENCP00000024751	AC012314.19
	AC012314.19
ENCP00000024811	LILRB4
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	LILRB4
ENCP00000024768	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024764	LILRA5
	LILRA5
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	LILRA5
	LILRA5
ENCP00000024756	LILRB2
	LILRB2
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ENCP00000024818	KIR2DP1

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ENCP00000024816	KIR3DL3
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ENCP00000024709	MYADM
	AC008753.6
ENCP00000024760	LILRA3
	LILRA3
	LILRA3
	LILRA3
ENCP00000024806	AC009892.10
	AC009892.10
ENCP00000024798	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024797	LILRA2
	LILRA2
	LILRA2
	LILRA2
ENCP00000024800	AC009892.5
ENCP00000024770	LAIR1
	LAIR1
	LAIR1

			LAIR1
			LAIR1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348830 (open chromatin region)	11	blood (K562)	H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC008984.5	downstream gene variant	541	ENST00000514037	?	?	4
AC008984.7	downstream gene variant	3024	ENST00000426257	?	?	3
AC010518.2	downstream gene variant, upstream gene variant	396	ENST00000615364	?	?	8
AC010518.3	downstream gene variant, upstream gene variant	54	ENST00000417373	?	?	23

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC008984.5	ENST00000514037	?	?	2