Locus 41 (rs2004318)

***SNiPA*card**

Block annotations

Block info

genomic range	chr19:55,117,749-55,168,602
block size	50,854 bp
variant count	74 variants

Basic features

Conservation/deleteriousness		**Linked genes**	
phyloP	μ = -0.557 [-4.065 – 2.368]	**gene(s) hit or close-by**	AC009892.10, AC009892.5, AC009892.9, AC245036.1, AC245036.2, AC245036.3, AC245036.4, AC245036.5, AC245036.6, LILRA1, LILRB1, LILRB4, MIR8061, VN1R105P
phastCons	μ = 0.059 [0 – 0.633]	**eQTL gene(s)**	CTB-83J4.2, LILRA1, LILRA2, LILRB2, LILRB5, LILRP1
GERP++	μ = -0.651 [-4.69 – 2.07]	**potentially regulated gene(s)**	AC008984.5, AC008984.5, AC008984.6, AC008984.6, AC008984.7, AC008984.7, AC009892.10, AC009892.10, AC009892.2, AC009892.2, AC009892.5, AC010518.3, AC010518.3, AC011515.2, AC011515.2, AC012314.19, AC012314.19, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, FCAR, KIR2DL1, KIR2DL3, KIR2DL4, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DP1, KIR2DS4, KIR3DL1, KIR3DL2, KIR3DL3, KIR3DX1, KIR3DX1, KIR3DX1, KIR3DX1, 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, LILRB4, LILRB4, LILRB4, LILRB4, LILRB4, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, VN1R105P
CADD score	μ = 3.422 [0.002 – 12.92]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
3-(4-hydroxyphenyl)lactate	3.53×10^{-5}	Metabolomics GWAS Server	24816252	3

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)
LILRB1	missense variant	ENST00000421584	?	ENSP00000410165	5	5			5
LILRB1	missense variant	ENST00000396315	?	ENSP00000379608	5	5			5
LILRB1	missense variant	ENST00000427581	?	ENSP00000395004	5	5			5
LILRB1	missense variant	ENST00000396332	NM_001081639.2	ENSP00000379623	5	5			5
LILRB1	missense variant	ENST00000396327	NM_001081638.2	ENSP00000379618	5	5			5
LILRB1	missense variant	ENST00000324602	NM_001081637.1	ENSP00000315997	5	5			5
LILRB1	missense variant	ENST00000396317	NM_001278398.1	ENSP00000379610	5	5			5
LILRB1	missense variant	ENST00000396331	NM_006669.5	ENSP00000379622	5	5			5
LILRB1	missense variant	ENST00000418536	?	ENSP00000391514	A/T	Gcc/Acc	?	?	1
LILRB1	missense variant	ENST00000396321	?	ENSP00000379614	A/T	Gcc/Acc	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRA1	?	ENSG00000104974	lung	4.15×10^{-7} (p-value)	GTEx Portal V6	39
LILRA2	?	ENSG00000239998	lung	1.26×10^{-7} (p-value)	GTEx Portal V6	61
LILRP1	?	ENSG00000186152	EBV lymphocytes	6.26×10^{-8} (p-value)	GTEx Portal V6	13
LILRB5	?	ENSG00000105609	EBV lymphocytes	1.02×10^{-7} (p-value)	GTEx Portal V6	51
LILRB2	?	ENSG00000131042	EBV lymphocytes	5.43×10^{-9} (p-value)	GTEx Portal V6	56
CTB-83J4.2	?	ENSG00000269271	blood	4.50×10^{-8} (p-value)	GTEx Portal V6	38
LILRA1	?	ENSG00000104974	blood	2.28×10^{-9} (p-value)	GTEx Portal V6	52
LILRA2	?	ENSG00000239998	subcutaneous adipocytes	1.86×10^{-8} (p-value)	GTEx Portal V6	5

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000344796	1	ENST00000418536, ENST00000396321	LILRB1

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000024800		

ENCP00000024805			LILRB1 LILRB1 LILRB1 LILRB1

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233609	1	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024761	AC008984.5 AC008984.5
		ENCP00000024823	KIR2DS4 KIR2DS4
		ENCP00000024769	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
		ENCP00000024758	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
		ENCP00000024812	AC011515.2 AC011515.2
		ENCP00000024753	LILRA6 LILRA6 LILRA6 LILRA6 LILRA6
		ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3

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ENCP00000024829	NLRP2
	NLRP2
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ENCP00000024788	LENG8
	LENG8
	LENG8
	LENG8
ENCP00000024803	LILRB1
	LILRB1
	LILRB1
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ENCP00000024773	LAIR1
	LAIR1
	LAIR1
	LAIR1
	LAIR1
ENCP00000024808	VN1R105P
ENCP00000024763	LILRA5
	AC008984.7
	AC008984.7
	LILRA5
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ENCP00000024821	KIR2DL4
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ENCP00000024772	LAIR1
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ENCP00000024825	FCAR
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ENCP00000024771	LAIR1
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ENCP00000024810	LILRB4
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ENCP00000024759	AC010518.3
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ENCP00000024819	KIR2DL1
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ENCP00000024799	LILRA1
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ENCP00000024751	AC012314.19
	AC012314.19
ENCP00000024811	LILRB4
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ENCP00000024768	LAIR1
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	LAIR1

ENCP00000024756	LILRB2 LILRB2 LILRB2 LILRB2 LILRB2
ENCP00000024764	LILRA5 LILRA5 LILRA5 LILRA5 LILRA5
ENCP00000024822	KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1 KIR3DL1
ENCP00000024818	KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1
ENCP00000024816	KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3 KIR3DL3

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		ENCP00000024760	LILRA3
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			LILRA3
			LILRA3
		ENCP00000024806	AC009892.10
			AC009892.10
		ENCP00000024798	LILRA2
			LILRA2
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		ENCP00000024797	LILRA2
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		ENCP00000024824	KIR3DL2
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		ENCP00000024800	AC009892.5
		ENCP00000024770	LAIR1
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			LAIR1
			LAIR1
			LAIR1
ENCE00000233672	1	ENCP00000024805	LILRB1
			LILRB1
			LILRB1
			LILRB1
			LILRB1
		ENCP00000024796	AC009892.2
			AC009892.2

		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233675	1	ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233680	3	ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024801	LILRB1

		LILRB1 LILRB1 LILRB1 LILRB1
	ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348881 (open chromatin region)	6	blood (GM12878)	H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000348884 (promoter)	3	cervix (HeLa-S3)	H3K27me3
		embryonic stem cell (H1ESC)	DNase1
		blood (GM12878)	Yy1, PU1, PolII, BATF, DNase1, IRF4, H3K79me2, H3K4me3, BCL11A, Pax5, H3K27ac, H3K4me2, H3K9ac
		A549	H3K27me3
		blood (K562)	H3K27me3
ENSR00001649225 (promoter)	1	cervix (HeLa-S3)	H3K27me3
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K4me2, H3K27ac
		blood (GM12878)	PolII, H3K36me3
		blood (K562)	H3K27me3
		blood (DND-41)	H3K27me3
		A549	H3K27me3
ENSR00000348890 (open chromatin region)	2	blood (K562)	USF1, H3K27me3
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC009892.10	downstream gene variant, upstream gene variant	347	ENST00000456337	?	?	14
AC009892.5	upstream gene variant, downstream gene variant	72	ENST00000432400	?	?	15
AC009892.9	upstream gene variant, downstream gene variant	2348	ENST00000429259	?	?	5
AC245036.1	downstream gene variant, upstream gene variant	20	ENST00000578908	?	?	22
AC245036.2	upstream gene variant, downstream gene variant	1613	ENST00000618840	?	?	4
AC245036.3	upstream gene variant, downstream gene variant	2000	ENST00000610849	?	?	4
AC245036.4	upstream gene variant, downstream gene variant	1785	ENST00000622837	?	?	4
AC245036.5	upstream gene variant, downstream gene variant	1871	ENST00000616955	?	?	4
AC245036.6	upstream gene variant, downstream gene variant	2086	ENST00000614474	?	?	4
LILRA1	downstream gene variant	4407	ENST00000473156	?	?	3
LILRA1	downstream gene variant	4194	ENST00000453777	NM_001278318.1	ENSP00000413715	4
LILRB1	upstream gene variant, downstream gene variant	195	ENST00000487425	?	?	22
LILRB1	upstream gene variant	225	ENST00000630741	?	ENSP00000485956	11
LILRB1	upstream gene variant	187	ENST00000418536	?	ENSP00000391514	11
LILRB1	upstream gene variant, downstream gene variant	37	ENST00000480375	?	?	20
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396315	?	ENSP00000379608	13
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396332	NM_001081639.2	ENSP00000379623	12
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000324602	NM_001081637.1	ENSP00000315997	12
LILRB1	upstream gene variant	225	ENST00000396321	?	ENSP00000379614	11
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396331	NM_006669.5	ENSP00000379622	6
LILRB1	upstream gene variant, downstream gene variant	613	ENST00000427581	?	ENSP00000395004	12
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396317	NM_001278308.1	ENSP00000379610	13

LILRB1	upstream gene variant, downstream gene variant	225	ENST00000396317	NM_001278398.1	ENSP00000379610	13
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000462628	?	?	10
LILRB1	upstream gene variant, downstream gene variant	477	ENST00000473412	?	?	16
LILRB1	upstream gene variant, downstream gene variant	275	ENST00000396327	NM_001081638.2	ENSP00000379618	12
LILRB1	upstream gene variant, downstream gene variant	225	ENST00000421584	?	ENSP00000410165	13
LILRB1	upstream gene variant, downstream gene variant	333	ENST00000480257	?	?	28
LILRB4	upstream gene variant, downstream gene variant	137	ENST00000461839	?	?	2
LILRB4	upstream gene variant	4977	ENST00000391736	NM_001278426.2	ENSP00000375616	1
LILRB4	upstream gene variant	3684	ENST00000270452	?	ENSP00000270452	1
MIR8061	downstream gene variant	1485	ENST00000618936	?	?	3
VN1R105P	upstream gene variant, downstream gene variant	117	ENST00000486595	?	?	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	2	3	3
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	2	3	3
LILRB1	ENST00000427581	?	ENSP00000395004	2	3	3
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	2	3	3
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	2	3	3
LILRB1	ENST00000421584	?	ENSP00000410165	2	2	3
LILRB1	ENST00000396315	?	ENSP00000379608	2	3	3
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	2	2	3

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000462628	?	?	1
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	1
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	1
LILRB1	ENST00000427581	?	ENSP00000395004	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	1
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	1
LILRB1	ENST00000421584	?	ENSP00000410165	1
LILRB1	ENST00000396315	?	ENSP00000379608	1
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC009892.10	ENST00000456337	?	?	7
LILRB1	ENST00000462628	?	?	15
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	39
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	14
LILRB1	ENST00000480257	?	?	1

LILRB1	ENST00000427581	?	ENSP00000395004	14
LILRB1	ENST00000480375	?	?	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	14
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	14
LILRB1	ENST00000421584	?	ENSP00000410165	13
LILRB1	ENST00000396315	?	ENSP00000379608	13
LILRB1	ENST00000396321	?	ENSP00000379614	25
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	13
LILRB1	ENST00000418536	?	ENSP00000391514	25
LILRB1	ENST00000487425	?	?	1
LILRB1	ENST00000630741	?	ENSP00000485956	26
LILRB4	ENST00000270452	?	ENSP00000270452	6
LILRB4	ENST00000461839	?	?	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000421584	?	ENSP00000410165	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRB1	ENST00000427581	?	ENSP00000395004	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC009892.5	ENST00000432400	?	3
LILRB1	ENST00000462628	?	13
LILRB1	ENST00000473412	?	8
LILRB4	ENST00000461839	?	1