Locus 125 (rs45461791)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,110,880-55,149,838
block size	38,959 bp
variant count	51 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.429 [-3.615 – 1.303]	gene(s) hit or close-by	AC009892.10, AC009892.5, AC245036.1, LILRA1, LILRB1
phastCons	μ = 0.067 [0 – 0.948]	eQTL gene(s)	LILRB1
GERP++	μ = -0.277 [-3.51 – 2.1]	potentially regulated gene(s)	AC008746.10, AC008746.10, AC008984.2, AC008984.2, AC008984.6, AC008984.6, AC009892.2, AC009892.2, AC009892.5, AC009892.9, AC012314.20, AC012314.20, AC012314.8, AC012314.8, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, AC098784.1, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, CNOT3, KIR3DX1, KIR3DX1, KIR3DX1, KIR3DX1, LAIR1, LAIR1, LAIR1, LAIR1, LAIR1, LAIR2, LAIR2, LAIR2, LAIR2, LILRA4, LILRA4, LILRA4, LILRA4, LILRB1, LILRB1, LILRB1, LILRB1, LILRB1, LILRP2, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, MBOAT7, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TMC4, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TSEN34, TTYH1, TTYH1, TTYH1, TTYH1
CADD score	μ = 3.491 [0.047 – 15.32]	disease gene(s)	, TSEN34, , , , , , , , , CNOT3

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
TSEN34	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	PONTOCEREBELLAR HYPOPLASIA, TYPE 2C	OMIM	MIM:612390
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
TSEN34	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970
	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970

	Pontocerebellar Hypoplasia Type 2 and Type 4	DECIPHER	MIM:316970	OMIM
TSEN34	Pontocerebellar hypoplasia type 2	OrphaNet	OrphaNet:2524	orphanet
CNOT3	Precursor T-cell acute lymphoblastic leukemia	OrphaNet	OrphaNet:99861	orphanet

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)
AC009892.5	splice donor variant	ENST00000432400	?	?					1
LILRB1	missense variant	ENST00000324602	NM_001081637.1	ENSP00000315997	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396315	?	ENSP00000379608	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396331	NM_006669.5	ENSP00000379622	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000427581	?	ENSP00000395004	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396327	NM_001081638.2	ENSP00000379618	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000421584	?	ENSP00000410165	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396317	NM_001278398.1	ENSP00000379610	V/L	Gtg/Ctg	?	?	1
LILRB1	missense variant	ENST00000396332	NM_001081639.2	ENSP00000379623	V/L	Gtg/Ctg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
LILRB1	ENST00000462628	ILMN_1708248	blood	9.02×10^{-31} (p-value)	MuTHER consortium	1
LILRB1	ENST00000396315					
LILRB1	ENST00000427581					
LILRB1	ENST00000396327					
LILRB1	ENST00000396317					
LILRB1	ENST00000480375					
LILRB1	ENST00000324602					
LILRB1	ENST00000421584					
LILRB1	ENST00000396332					
LILRB1	ENST00000396331					
LILRB1	?	ENSG00000104972	testis	3.99×10^{-6} (p-value)	GTEx Portal V6	1

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		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233565	2	ENCP00000024749	AC012314.20 AC012314.20
		ENCP00000024780	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024781	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024748	TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34 TSEN34
		ENCP00000024778	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024742	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024745	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024733	CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3 CNOT3
		ENCP00000024804	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024777	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024741	TMC4 TMC4 TMC4

			TMC4 TMC4 TMC4 TMC4 TMC4 TMC4 TMC4
		ENCP00000024728	AC012314.8 AC012314.8
ENCE00000233676	1	ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
		ENCP00000024826	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024805	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024809	AC009892.9
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024793	LAIR2 LAIR2 LAIR2 LAIR2
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCE00000233601	1	ENCP00000024767	LILRA4 LILRA4 LILRA4 LILRA4 AC008984.2 AC008984.2
		ENCP00000024744	MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7 MBOAT7
		ENCP00000024775	AC008746.10 AC008746.10
		ENCP00000024779	TTYH1 TTYH1 TTYH1 TTYH1
		ENCP00000024792	LAIR2 LAIR2 LAIR2 LAIR2

		ENCP00000024814	AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1 AC098784.1
		ENCP00000024815	LILRP2 LILRP2
		ENCP00000024774	LAIR1 LAIR1 LAIR1 LAIR1 LAIR1
ENCE00000233679	1	ENCP00000024815	LILRP2 LILRP2
		ENCP00000024796	AC009892.2 AC009892.2
		ENCP00000024766	AC008984.6 AC008984.6
		ENCP00000024793	LAIR2 LAIR2 LAIR2

		ENCP00000024826	LAIR2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
		ENCP00000024801	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024795	KIR3DX1 KIR3DX1 KIR3DX1 KIR3DX1
ENCE00000233738	1	ENCP00000024793	LAIR2 LAIR2 LAIR2 LAIR2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348881 (open chromatin region)	7	blood (GM12878)	H3K4me3, H3K27ac, H3K4me2
		blood (K562)	H3K27me3
		A549	H3K27me3
ENSR00000348884 (promoter)	1	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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC009892.10	downstream gene variant	1067	ENST00000456337	?	?	3
AC009892.5	upstream gene variant, downstream gene variant	53	ENST00000432400	?	?	32
AC245036.1	downstream gene variant, upstream gene variant	678	ENST00000578908	?	?	5
LILRA1	downstream gene variant	1211	ENST00000495417	?	?	2
LILRA1	downstream gene variant	1447	ENST00000618665	?	ENSP00000479482	2
LILRA1	downstream gene variant	2156	ENST00000453777	NM_001278318.1	ENSP00000413715	4
LILRA1	downstream gene variant	1215	ENST00000477255	?	?	2
LILRA1	downstream gene variant	3171	ENST00000251372	NM_006863.3	ENSP00000251372	3
LILRA1	downstream gene variant	2369	ENST00000473156	?	?	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000324602	NM_001081637.1	ENSP00000315997	3
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000421584	?	ENSP00000410165	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396331	NM_006669.5	ENSP00000379622	4
LILRB1	upstream gene variant	2360	ENST00000396321	?	ENSP00000379614	2
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000396317	NM_001278398.1	ENSP00000379610	4
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000462628	?	?	2
LILRB1	upstream gene variant	2360	ENST00000630741	?	ENSP00000485956	2
LILRB1	upstream gene variant, downstream gene variant	238	ENST00000480375	?	?	6

LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396332	NM_001081639.2	ENSP00000379623	3
LILRB1	upstream gene variant, downstream gene variant	859	ENST00000396327	NM_001081638.2	ENSP00000379618	4
LILRB1	upstream gene variant, downstream gene variant	2230	ENST00000480257	?	?	6
LILRB1	upstream gene variant, downstream gene variant	315	ENST00000396315	?	ENSP00000379608	4
LILRB1	upstream gene variant, downstream gene variant	102	ENST00000473412	?	?	7
LILRB1	upstream gene variant, downstream gene variant	201	ENST00000427581	?	ENSP00000395004	4
LILRB1	upstream gene variant, downstream gene variant	86	ENST00000487425	?	?	4
LILRB1	upstream gene variant	2322	ENST00000418536	?	ENSP00000391514	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
AC009892.10	ENST00000456337	?	?	3
LILRA1	ENST00000251372	NM_006863.3	ENSP00000251372	1
LILRA1	ENST00000453777	NM_001278318.1	ENSP00000413715	1
LILRA1	ENST00000473156	?	?	1
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	3
LILRB1	ENST00000396321	?	ENSP00000379614	6
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	4
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	4
LILRB1	ENST00000630741	?	ENSP00000485956	6
LILRB1	ENST00000418536	?	ENSP00000391514	6
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	12
LILRB1	ENST00000421584	?	ENSP00000410165	3
LILRB1	ENST00000396315	?	ENSP00000379608	3
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	4
LILRB1	ENST00000462628	?	?	5
LILRB1	ENST00000427581	?	ENSP00000395004	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LILRA1	ENST00000618665	?	ENSP00000479482	1
LILRA1	ENST00000251372	NM_006863.3	ENSP00000251372	2
LILRA1	ENST00000453777	NM_001278318.1	ENSP00000413715	2
LILRB1	ENST00000396317	NM_001278398.1	ENSP00000379610	1
LILRB1	ENST00000324602	NM_001081637.1	ENSP00000315997	2
LILRB1	ENST00000396327	NM_001081638.2	ENSP00000379618	2
LILRB1	ENST00000396331	NM_006669.5	ENSP00000379622	2
LILRB1	ENST00000421584	?	ENSP00000410165	1
LILRB1	ENST00000396315	?	ENSP00000379608	1
LILRB1	ENST00000396332	NM_001081639.2	ENSP00000379623	2
LILRB1	ENST00000427581	?	ENSP00000395004	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
AC009892.5	ENST00000432400	?	1
LILRA1	ENST00000477255	?	1
LILRA1	ENST00000495417	?	1
LILRA1	ENST00000473156	?	2
LILRB1	ENST00000462628	?	4
LILRB1	ENST00000487425	?	2