Locus 355 (rs11672111)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,562,797-55,566,258
block size	3,462 bp
variant count	22 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.703 [-2.297 – 0.425]	**gene(s) hit or close-by**	CTC-550B14.6, CTC-550B14.7, RDH13
phastCons	μ = 0.016 [0 – 0.209]	**eQTL gene(s)**	GP6
GERP++	μ = 0.034 [-0.765 – 0.547]	**potentially regulated gene(s)**	AC009892.10, AC009892.10, AC009892.5, AC011515.2, AC011515.2, CTC-550B14.1, 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, LILRA1, LILRA1, LILRA1, LILRA1, LILRA2, LILRA2, LILRA2, LILRA2, LILRB1, LILRB1, LILRB1, LILRB1, LILRB1, LILRB4, LILRB4, LILRB4, LILRB4, LILRB4, LILRP1, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, NLRP2, VN1R105P
CADD score	μ = 3.874 [0.138 – 10.17]	**disease gene(s)**	GP6

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
GP6	BLEEDING DISORDER, PLATELET-TYPE, 11	OMIM	MIM:614201 OMIM®
GP6	Bleeding diathesis due to glycoprotein VI deficiency	OrphaNet	OrphaNet:98885 orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
GP6	?	ENSG00000088053	sun exposed skin	1.22×10^{-7} (p-value)	GTEx Portal V6	21
GP6	?	ENSG00000088053	unexposed skin	1.93×10^{-9} (p-value)	GTEx Portal V6	21

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000233760	1	ENCP00000024802	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
		ENCP00000024807	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
		ENCP00000024823	KIR2DS4 KIR2DS4
		ENCP00000024813	LILRP1 LILRP1

ENCP00000024799	LILRA1 LILRA1 LILRA1 LILRA1
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024811	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
ENCP00000024829	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
ENCP00000024822	KIR3DL1 KIR3DL1
ENCP00000024818	KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1 KIR2DP1

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ENCP00000024816	KIR3DL3
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ENCP00000024803	LILRB1
	LILRB1
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	LILRB1
ENCP00000024808	VN1R105P
ENCP00000024798	LILRA2
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ENCP00000024806	AC009892.10
	AC009892.10
ENCP00000024824	KIR3DL2
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ENCP00000024797	LILRA2
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	LILRA2
ENCP00000024800	AC009892.5
ENCP00000024821	KIR2DL4
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ENCP00000024825	FCAR
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ENCP00000024810	LILRB4
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ENCP00000024819	KIR2DL1
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		ENCP00000024807	LILRB4
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		ENCP00000024823	KIR2DS4
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		ENCP00000024813	LILRP1
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ENCP00000024799	LILRA1 LILRA1 LILRA1 LILRA1
ENCP00000024812	AC011515.2 AC011515.2
ENCP00000024817	KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3 KIR2DL3
ENCP00000024811	LILRB4 LILRB4 LILRB4 LILRB4 LILRB4
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

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ENCP00000024803	LILRB1 LILRB1 LILRB1 LILRB1 LILRB1
ENCP00000024827	NLRP2 NLRP2 NLRP2 NLRP2 NLRP2 CTC-550B14.1 NLRP2 NLRP2 NLRP2 NLRP2 NLRP2
ENCP00000024808	VN1R105P
ENCP00000024798	LILRA2 LILRA2 LILRA2 LILRA2
ENCP00000024806	AC009892.10 AC009892.10
ENCP00000024797	LILRA2 LILRA2 LILRA2 LILRA2
ENCP00000024824	KIR3DL2 KIR3DL2
ENCP00000024800	AC009892.5
ENCP00000024821	KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4 KIR2DL4

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ENCP00000024825	FCAR
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ENCP00000024810	LILRB4
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ENCP00000024819	KIR2DL1
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Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	upstream gene variant	56	ENST00000585492	?	?	9
CTC-550B14.7	downstream gene variant	3343	ENST00000593060	?	?	3
RDH13	downstream gene variant	1763	ENST00000586945	?	ENSP00000466708	21
RDH13	downstream gene variant	4363	ENST00000587026	?	ENSP00000467754	8

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	ENST00000585492	?	?	13
RDH13	ENST00000589305	?	?	22
RDH13	ENST00000591960	?	ENSP00000468422	22
RDH13	ENST00000291892	?	ENSP00000291892	22
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	22
RDH13	ENST00000587721	?	ENSP00000467208	22
RDH13	ENST00000592573	?	ENSP00000465538	22
RDH13	ENST00000589605	?	?	22
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	22
RDH13	ENST00000610356	?	ENSP00000477732	22

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTC-550B14.6	ENST00000585492	?	1