Locus 126 (rs1654439)

SNiPAcard

Block annotations

Block info

genomic range	chr19:55,548,269-55,571,055
block size	22,787 bp
variant count	16 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.333 [-2.99 – 0.001]	gene(s) hit or close-by	CTC-550B14.6, CTC-550B14.7, GP6, RDH13
phastCons	μ = 0.015 [0 – 0.135]	eQTL gene(s)	CTC-550B14.6, CTC-550B14.7, GP6, NLRP2, RDH13
GERP++	μ = -1.023 [-10.2 – 0.367]	potentially regulated gene(s)	–
CADD score	μ = 2.601 [0.974 – 5.875]	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
GP6	Bleeding diathesis due to glycoprotein VI deficiency	OrphaNet	OrphaNet:98885

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NLRP2	?	ENSG00000022556	visceral adipocytes	6.13×10-7 (p-value)	GTEx Portal V6	14
CTC-550B14.7	?	ENSG00000267265	lung	1.01×10-10 (p-value)	GTEx Portal V6	16
CTC-550B14.7	?	ENSG00000267265	spleen	8.64×10-11 (p-value)	GTEx Portal V6	14
RDH13	?	ENSG00000160439	spleen	2.66×10-6 (p-value)	GTEx Portal V6	2
NLRP2	?	ENSG00000022556	spleen	1.02×10-7 (p-value)	GTEx Portal V6	3
RDH13	?	ENSG00000160439	blood	1.40×10-7 (p-value)	GTEx Portal V6	7
CTC-550B14.7	?	ENSG00000267265	blood	8.09×10-8 (p-value)	GTEx Portal V6	14
NLRP2	?	ENSG00000022556	blood	6.52×10-8 (p-value)	GTEx Portal V6	12
CTC-550B14.6	?	ENSG00000267149	esophagus mucosa	2.71×10-8 (p-value)	GTEx Portal V6	14
NLRP2	?	ENSG00000022556	esophagus mucosa	6.53×10-6 (p-value)	GTEx Portal V6	1
GP6	?	ENSG00000088053	thyroid	3.22×10-6 (p-value)	GTEx Portal V6	6
NLRP2	?	ENSG00000022556	thyroid	1.73×10-6 (p-value)	GTEx Portal V6	2
CTC-550B14.6	?	ENSG00000267149	skeletal muscle	1.61×10-6 (p-value)	GTEx Portal V6	5
CTC-550B14.6	?	ENSG00000267149	sun exposed skin	2.53×10-20 (p-value)	GTEx Portal V6	16
RDH13	?	ENSG00000160439	sun exposed skin	2.71×10-13 (p-value)	GTEx Portal V6	16
CTC-550B14.6	?	ENSG00000267149	unexposed skin	7.77×10-13 (p-value)	GTEx Portal V6	16
RDH13	?	ENSG00000160439	unexposed skin	2.76×10-6 (p-value)	GTEx Portal V6	6

CTC-550B14.7	?	ENSG00000267265	subcutaneous adipocytes	2.24×10^{-8} (p-value)	GTEx Portal V6	16
NLRP2	ENST00000537859	ILMN_1664878	skin	4.74×10^{-13} (p-value)	MuTHER consortium	8
NLRP2	ENST00000540597		blood	1.39×10^{-20} (p-value)	MuTHER consortium	8
NLRP2	ENST00000263437					
NLRP2	ENST00000391721					
NLRP2	ENST00000339757					
NLRP2	ENST00000543010					
NLRP2	ENST00000448584					
NLRP2	ENST00000427260					
NLRP2	ENST00000542755					
NLRP2	?	ENSG00000022556	blood	1.37×10^{-3} (q-value)	SeeQTL DB (HapMap)	5
NLRP2	ENST00000537859	ILMN_1664878	monocyte	2.07×10^{-19} (p-value)	Zeller et al.	3
NLRP2	ENST00000540597					
NLRP2	ENST00000263437					
NLRP2	ENST00000391721					
NLRP2	ENST00000339757					
NLRP2	ENST00000543010					
NLRP2	ENST00000448584					
NLRP2	ENST00000427260					
NLRP2	ENST00000542755					
NLRP2	?	ENSG00000022556	tibial nerve	8.87×10^{-6} (p-value)	GTEx Portal V6	1
CTC-550B14.6	?	ENSG00000267149	stomach	8.27×10^{-7} (p-value)	GTEx Portal V6	3

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000348908 (CTCF binding site)	1	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, CTCF, Rad21
		blood (K562)	H3K36me3
		cervix (HeLa-S3)	CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	upstream gene variant	1158	ENST00000585492	?	?	1
CTC-550B14.7	downstream gene variant	92	ENST00000586845	?	?	5
CTC-550B14.7	downstream gene variant	1339	ENST00000586961	?	?	1
CTC-550B14.7	downstream gene variant	2116	ENST00000593060	?	?	2
GP6	upstream gene variant	648	ENST00000333884	NM_001256017.2	ENSP00000334552	5
GP6	upstream gene variant	648	ENST00000310373	NM_001083899.2	ENSP00000308782	5
GP6	upstream gene variant	648	ENST00000417454	NM_016363.5	ENSP00000394922	5
GP6	upstream gene variant	4036	ENST00000468330	?	ENSP00000464922	1

GP6	upstream gene variant	4036	ENST00000468239	?	ENSP00000464922	1
RDH13	downstream gene variant, upstream gene variant	164	ENST00000592423	?	?	8
RDH13	downstream gene variant, upstream gene variant	707	ENST00000591868	?	?	4
RDH13	downstream gene variant	3584	ENST00000591023	?	?	2
RDH13	downstream gene variant	47	ENST00000587026	?	ENSP00000467754	2
RDH13	downstream gene variant	4214	ENST00000587373	?	?	2
RDH13	downstream gene variant	3533	ENST00000588306	?	?	2
RDH13	downstream gene variant	4353	ENST00000587046	?	?	2
RDH13	downstream gene variant	331	ENST00000396247	NM_138412.3	ENSP00000379547	7
RDH13	downstream gene variant	843	ENST00000592573	?	ENSP00000465538	7
RDH13	downstream gene variant	672	ENST00000589305	?	?	1
RDH13	downstream gene variant	331	ENST00000291892	?	ENSP00000291892	7
RDH13	downstream gene variant	1567	ENST00000586945	?	ENSP00000466708	1
RDH13	downstream gene variant	331	ENST00000415061	NM_001145971.1	ENSP00000391121	7
RDH13	downstream gene variant	167	ENST00000591960	?	ENSP00000468422	3
RDH13	downstream gene variant	312	ENST00000610356	?	ENSP00000477732	7
RDH13	downstream gene variant	3821	ENST00000589197	?	?	2
RDH13	downstream gene variant	1757	ENST00000587721	?	ENSP00000467208	6
RDH13	downstream gene variant	4261	ENST00000593134	?	?	2
RDH13	downstream gene variant, upstream gene variant	167	ENST00000586331	?	?	10
RDH13	downstream gene variant	4065	ENST00000590349	?	?	2
RDH13	downstream gene variant, upstream gene variant	32	ENST00000589605	?	?	6
RDH13	downstream gene variant	3354	ENST00000591603	?	?	2

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
RDH13	ENST00000291892	?	ENSP00000291892	G	ggT/ggC	1
RDH13	ENST00000592573	?	ENSP00000465538	G	ggT/ggC	1
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	G	ggT/ggC	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CTC-550B14.6	ENST00000585492	?	?	13
CTC-550B14.7	ENST00000593060	?	?	11
CTC-550B14.7	ENST00000586845	?	?	6
CTC-550B14.7	ENST00000586961	?	?	10
GP6	ENST00000310373	NM_001083899.2	ENSP00000308782	1
GP6	ENST00000417454	NM_016363.5	ENSP00000394922	1
GP6	ENST00000333884	NM_001256017.2	ENSP00000334552	1
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	6
RDH13	ENST00000589605	?	?	3

RDH13	ENST00000586945	?	ENSP00000466708	2
RDH13	ENST00000291892	?	ENSP00000291892	6
RDH13	ENST00000610356	?	ENSP00000477732	6
RDH13	ENST00000592573	?	ENSP00000465538	6
RDH13	ENST00000415061	NM_001145971.1	ENSP00000391121	6
RDH13	ENST00000591960	?	ENSP00000468422	13
RDH13	ENST00000587721	?	ENSP00000467208	5
RDH13	ENST00000589305	?	?	4
RDH13	ENST00000591868	?	?	6
RDH13	ENST00000587026	?	ENSP00000467754	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
RDH13	ENST00000396247	NM_138412.3	ENSP00000379547	1
RDH13	ENST00000586945	?	ENSP00000466708	1
RDH13	ENST00000610356	?	ENSP00000477732	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CTC-550B14.7	ENST00000593060	?	3
CTC-550B14.7	ENST00000586845	?	3
CTC-550B14.7	ENST00000586961	?	3
RDH13	ENST00000586331	?	1
RDH13	ENST00000592423	?	3
RDH13	ENST00000589305	?	1