Locus 179 (rs2523570)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,329,734-31,333,914
block size	4,181 bp
variant count	11 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.707 [-2.823 – 0.83]	**gene(s) hit or close-by**	DHFRP2, HLA-B, HLA-C, RNU6-283P, XXbac-BPG248L24.12
phastCons	μ = 0.042 [0 – 0.185]	**eQTL gene(s)**	FLOT1, HCG22, HCP5, HLA-B, SKIV2L, XXbac-BPG248L24.12
GERP++	μ = -0.072 [-1.92 – 0.83]	**potentially regulated gene(s)**	CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, CSNK2B, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDR1, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, DDX39B, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GPANK1, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, GTF2H4, HCP5, HCP5, HCP5, HCP5, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-B, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, HLA-C, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LST1, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, LY6G5C, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, MICB, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PRRC2A, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, PSORS1C1, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, TNF, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, UQCRHP1, VARS, VARS, VARS, VARS, VARS, VARS, VARS, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, VARS2, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.10, XXbac-BPG248L24.12
CADD score	μ = 2.437 [0.078 – 4.885]	**disease gene(s)**	HLA-B, SKIV2L, , , , , , , TNF, , , , , , VARS2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
HLA-B	Takayasu arteritis	OrphaNet	OrphaNet:3287
HLA-B	Stevens-Johnson syndrome	OrphaNet	OrphaNet:36426
HLA-B	Behcet disease	OrphaNet	OrphaNet:117
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	PSORIASIS 1, SUSCEPTIBILITY TO	OMIM	MIM:177900
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507
TNF	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424
TNF	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162

TNF	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TOTOXIC EPIDERMAL [...]	OMIM	MIM:608579	OMIM
	HEPATITIS B VIRUS, SUSCEPTIBILITY TO	OMIM	MIM:610424	OMIM
	MALARIA, SUSCEPTIBILITY TOMALARIA, RESISTANCE TO, INCLUDED	OMIM	MIM:611162	OMIM
	PSORIATIC ARTHRITIS, SUSCEPTIBILITY TOPSORIATIC ARTHRITIS, SUSCEPTIBILITY [...]	OMIM	MIM:607507	OMIM
VARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 20	OMIM	MIM:615917	OMIM
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
XXbac-BPG248L24.12	?	ENSG00000271581	pancreas	1.22×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	muscularis mucosae	8.92×10-12 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	lung	4.02×10-13 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	transformed fibroblasts	1.05×10-10 (p-value)	GTEx Portal V6	6
HLA-B	?	ENSG00000234745	blood	2.32×10-7 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	blood	3.21×10-18 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	breast	1.65×10-14 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	tibial artery	2.50×10-11 (p-value)	GTEx Portal V6	11
FLOT1	?	ENSG00000137312	thyroid	3.53×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	thyroid	7.04×10-11 (p-value)	GTEx Portal V6	11
HLA-B	?	ENSG00000234745	skeletal muscle	1.41×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	skeletal muscle	1.03×10-18 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	EBV lymphocytes	9.39×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	transverse colon	1.80×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	liver	2.78×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	sun exposed skin	1.94×10-12 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	unexposed skin	5.48×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	aorta	1.88×10-10 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	left ventricle	1.15×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	subcutaneous adipocytes	7.46×10-13 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	visceral adipocytes	5.61×10-10 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	cortex	1.69×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	tibial nerve	2.22×10-10 (p-value)	GTEx Portal V6	11

XXbac-BPG248L24.12	?	ENSG00000271581	esophagus mucosa	2.56×10-11 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	gastroesophageal junction	4.28×10-11 (p-value)	GTEx Portal V6	11
XXbac-BPG248L24.12	?	ENSG00000271581	adrenal gland	4.12×10-6 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	testis	4.56×10-7 (p-value)	GTEx Portal V6	1
XXbac-BPG248L24.12	?	ENSG00000271581	frontal cortex	1.01×10-6 (p-value)	GTEx Portal V6	1
HCG22	ENST00000570223	ILMN_1667229	blood	3.80×10-5 (p-value)	MuTHER consortium	2
HCG22	ENST00000565192					
HCG22	ENST00000615046					
HCG22	ENST00000426185					
HCG22	ENST00000562344					
SKIV2L	ENST00000484835	ILMN_1666512	monocyte	3.43×10-4 (p-value)	Fairfax et al.	1
SKIV2L	ENST00000375394					
SKIV2L	ENST00000474839					
SKIV2L	ENST00000470453					
SKIV2L	ENST00000465703					
SKIV2L	ENST00000491994					
SKIV2L	ENST00000471818					
SKIV2L	ENST00000483553					
SKIV2L	ENST00000485349					
?	?	1557242_at	blood	1.30×10-10 (p-value)	Dixon et al.	1
HCP5	ENST00000414046	ILMN_1803945	blood	7.20×10-7 (p-value)	Westra et al.	1
HCP5	ENST00000541196					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444665	1	ENCP00000048065	CSNK2B GPANK1 GPANK1 GPANK1 CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B CSNK2B GPANK1 GPANK1 GPANK1 GPANK1
		ENCP00000048069	LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C LY6G5C
		ENCP00000048002	XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10 XXbac-BPG248L24.10

ENCP00000048005	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048017	HCP5 HCP5 HCP5 HCP5
ENCP00000048050	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000047997	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048023	DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B DDX39B
ENCP00000048104	VARS VARS VARS VARS VARS VARS VARS
ENCP00000047959	DDR1 DDR1 DDR1 DDR1 DDR1 DDR1 DDR1
ENCP00000047963	GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4 GTF2H4
ENCP00000048052	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048110	VARS VARS VARS VARS VARS VARS VARS
ENCP00000048109	VARS VARS VARS VARS

	VARS VARS VARS
ENCP00000048051	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048003	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048033	TNF TNF TNF TNF TNF TNF TNF
ENCP00000047998	HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C HLA-C
ENCP00000048035	LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048004	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048007	HLA-B HLA-B HLA-B HLA-B HLA-B HLA-B
ENCP00000048048	PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A PRRC2A
ENCP00000048063	GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1 GPANK1
ENCP00000047979	PSORS1C1 PSORS1C1 PSORS1C1 PSORS1C1 PSORS1C1

	PSORS1C1
ENCP00000048006	XXbac-BPG248L24.12
ENCP00000048040	UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1 UQCRHP1
ENCP00000048037	LST1 LST1 LST1 LST1 LST1 LST1 LST1 LST1
ENCP00000048020	MICB MICB MICB MICB MICB MICB MICB MICB
ENCP00000047966	VARS2 VARS2 VARS2 VARS2 VARS2 VARS2

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHFRP2	downstream gene variant	215	ENST00000414224	?	?	11
HLA-B	upstream gene variant	4770	ENST00000412585	NM_005514.6	ENSP00000399168	1
HLA-B	upstream gene variant	4778	ENST00000498007	?	?	1
HLA-B	upstream gene variant	4769	ENST00000434333	?	ENSP00000405931	1
HLA-C	upstream gene variant	4799	ENST00000620806	?	ENSP00000480331	1
RNU6-283P	upstream gene variant	3997	ENST00000364788	?	?	4
XXbac-BPG248L24.12	downstream gene variant	4320	ENST00000603274	?	?	1