Locus 123 (rs9283893)

SNiPAcard

Block annotations

Block info

genomic range	chr6:31,882,421-31,935,928
block size	53,508 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.590 [-1.89 – 0.405]	gene(s) hit or close-by	C2, DXO, MIR1236, NELFE, SKIV2L, STK19, XXbac-BPG116M5.17
phastCons	μ = 0.025 [0 – 0.14]	eQTL gene(s)	SKIV2L
GERP++	μ = 0.497 [-0.607 – 1.57]	potentially regulated gene(s)	–
CADD score	μ = 3.537 [0.258 – 7.157]	disease gene(s)	SKIV2L, C2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	DECIPHER	MIM:615602
SKIV2L	TRICHOHEPATOENTERIC SYNDROME 2	OMIM	MIM:614602
C2	COMPLEMENT COMPONENT 2 DEFICIENCY	OMIM	MIM:217000
C2	MACULAR DEGENERATION, AGE-RELATED, 1	OMIM	MIM:603075
SKIV2L	Syndromic diarrhea	OrphaNet	OrphaNet:84064
C2	Immunodeficiency due to an early component of complement deficiency	OrphaNet	OrphaNet:169147

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SKIV2L	?	ENSG00000204351	muscularis mucosae	3.96×10-6 (p-value)	GTEx Portal V6	1
SKIV2L	?	ENSG00000204351	tibial nerve	6.19×10-7 (p-value)	GTEx Portal V6	3
SKIV2L	?	ENSG00000204351	transverse colon	9.58×10-8 (p-value)	GTEx Portal V6	3
SKIV2L	?	ENSG00000204351	unexposed skin	1.40×10-8 (p-value)	GTEx Portal V6	5
SKIV2L	?	ENSG00000204351	tibial artery	4.93×10-8 (p-value)	GTEx Portal V6	4
SKIV2L	?	ENSG00000204351	thyroid	1.77×10-7 (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494593 (enhancer)	1	liver (HepG2)	H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K79me2, ELF1, H3K4me1
		HSMMtube	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
C2	upstream gene variant	2906	ENST00000494905	?	ENSP00000419048	1
C2	upstream gene variant	2111	ENST00000411571	?	ENSP00000388727	2
C2	upstream gene variant, downstream gene variant	548	ENST00000413154	?	ENSP00000403325	3
C2	upstream gene variant	2266	ENST00000484636	?	ENSP00000420305	2
C2	upstream gene variant	4841	ENST00000485690	?	ENSP00000417790	1
DXO	downstream gene variant	1659	ENST00000477826	?	?	1
DXO	downstream gene variant	1693	ENST00000375356	?	ENSP00000364505	1
DXO	downstream gene variant	1666	ENST00000478221	?	?	1
DXO	downstream gene variant	1659	ENST00000491327	?	?	1
DXO	downstream gene variant	2492	ENST00000487914	?	?	1
DXO	downstream gene variant	2291	ENST00000492946	?	?	1
DXO	downstream gene variant	1662	ENST00000375349	?	ENSP00000364498	1
DXO	downstream gene variant	1797	ENST00000474587	?	?	1
DXO	downstream gene variant	1756	ENST00000480240	?	ENSP00000418632	1
DXO	downstream gene variant	1736	ENST00000498357	?	?	1
DXO	downstream gene variant	1659	ENST00000337523	NM_005510.3	ENSP00000337759	1
DXO	downstream gene variant	1664	ENST00000495340	?	ENSP00000419588	1
DXO	downstream gene variant	1659	ENST00000485557	?	?	1
DXO	downstream gene variant	1664	ENST00000473976	?	?	1
DXO	downstream gene variant	2747	ENST00000460058	?	?	1
MIR1236	upstream gene variant	980	ENST00000408340	?	?	1
SKIV2L	upstream gene variant	3577	ENST00000466290	?	?	1
SKIV2L	upstream gene variant	373	ENST00000470453	?	?	1
SKIV2L	upstream gene variant	324	ENST00000491994	?	ENSP00000417586	1
SKIV2L	upstream gene variant	1271	ENST00000492900	?	?	1
SKIV2L	upstream gene variant	1197	ENST00000488648	?	?	1
SKIV2L	upstream gene variant	1251	ENST00000474839	?	ENSP00000420470	1
SKIV2L	upstream gene variant	1160	ENST00000375394	NM_006929.4	ENSP00000364543	1
SKIV2L	upstream gene variant	1221	ENST00000465703	?	?	1
SKIV2L	upstream gene variant	1237	ENST00000461073	?	ENSP00000419905	1
SKIV2L	upstream gene variant	1273	ENST00000628157	?	ENSP00000485707	1
SKIV2L	upstream gene variant	172	ENST00000471818	?	?	1
SKIV2L	upstream gene variant	303	ENST00000485349	?	?	1
STK19	upstream gene variant	3793	ENST00000375333	NM_032454.1	ENSP00000364482	1
STK19	upstream gene variant	4189	ENST00000473983	?	?	1
STK19	upstream gene variant	3015	ENST00000460018	?	ENSP00000418350	1
STK19	upstream gene variant	4431	ENST00000469907	?	?	1
STK19	upstream gene variant	4230	ENST00000483801	?	ENSP00000418866	1
STK19	upstream gene variant	3680	ENST00000375331	NM_004197.1	ENSP00000364480	1
STK19	upstream gene variant	3680	ENST00000479644	?	?	1
STK19	upstream gene variant	3748	ENST00000492583	?	?	1

STK19	upstream gene variant	4427	ENST00000484540	?	?	1
STK19	upstream gene variant	4223	ENST00000463823	?	?	1
STK19	upstream gene variant	4384	ENST00000471028	?	?	1
STK19	upstream gene variant	4230	ENST00000519179	?	ENSP00000454870	1
STK19	upstream gene variant	4246	ENST00000466336	?	?	1
STK19	upstream gene variant	2940	ENST00000466132	?	?	1
STK19	upstream gene variant	4201	ENST00000478486	?	?	1
STK19	upstream gene variant	4404	ENST00000491861	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C2	ENST00000484636	?	ENSP00000420305	1
C2	ENST00000299367	NM_001282458.1, NM_000063.5	ENSP00000299367	1
C2	ENST00000452323	NM_001178063.2	ENSP00000392322	4
C2	ENST00000442278	NM_001145903.2	ENSP00000395683	1
C2	ENST00000452202	?	ENSP00000406121	4
C2	ENST00000482060	?	ENSP00000418332	1
C2	ENST00000469372	NM_001282457.1	ENSP00000418923	4
C2	ENST00000418949	NM_001282459.1	ENSP00000406190	1
C2	ENST00000497706	?	ENSP00000417482	4
C2	ENST00000383177	?	ENSP00000372664	1
C2	ENST00000411571	?	ENSP00000388727	1
C2	ENST00000447952	?	ENSP00000391354	1
NELFE	ENST00000491139	?	?	1
NELFE	ENST00000436289	?	ENSP00000414029	1
NELFE	ENST00000492185	?	?	1
NELFE	ENST00000481121	?	?	1
NELFE	ENST00000426722	?	ENSP00000394340	1
NELFE	ENST00000375429	NM_002904.5	ENSP00000364578	1
NELFE	ENST00000625905	?	ENSP00000486884	1
NELFE	ENST00000375425	?	ENSP00000364574	1
NELFE	ENST00000444811	?	ENSP00000388400	1
NELFE	ENST00000492539	?	ENSP00000419183	1
NELFE	ENST00000441998	?	ENSP00000397914	1
NELFE	ENST00000494956	?	ENSP00000420205	1
NELFE	ENST00000488426	?	?	1
NELFE	ENST00000454913	?	ENSP00000409389	1
SKIV2L	ENST00000484835	?	?	1
SKIV2L	ENST00000375394	NM_006929.4	ENSP00000364543	1

SKIV2L	ENST00000461915	?	?	1
SKIV2L	ENST00000474839	?	ENSP00000420470	1
SKIV2L	ENST00000483553	?	ENSP00000420332	1
SKIV2L	ENST00000465703	?	?	1
XXbac-BPG116M5.17	ENST00000477310	?	ENSP00000418996	1
XXbac-BPG116M5.17	ENST00000456570	?	ENSP00000410815	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
SKIV2L	ENST00000494058	?	1