SNiPAcard

Block annotations

Block info

genomic range	chr1:157,661,848-157,693,722
block size	31,875 bp
variant count	13 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.795 [-3.247 – 0.025]	**gene(s) hit or close-by**	FCRL3, RP11-367J7.3, VDAC1P9
phastCons	μ = 0.062 [0 – 0.694]	**eQTL gene(s)**	ATP1A4, FCRL1, FCRL3, RP11-367J7.3
GERP++	μ = -1.674 [-10.7 – 1.7]	**potentially regulated gene(s)**	–
CADD score	μ = 2.100 [0.524 – 4.934]	**disease gene(s)**	FCRL3

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Graves' disease	$<2.00\times10^{-13}$	GWAS Catalog	21841780		1
Multiple sclerosis	$<3.00\times10^{-6}$	GWAS Catalog	21833088		1
Type 1 diabetes autoantibodies	$<1.00\times10^{-11}$	GWAS Catalog	21829393		1
Alkaline phosphatase	8.18×10^{-5}	dbGaP	pha001181	dbGaP	1
Alkaline phosphatase	3.25×10^{-5}	dbGaP	pha001179	dbGaP	1

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
?	HGMD curated	HGMD	CR067135	HGMD®	1
?	HGMD curated	HGMD	CR067134	HGMD®	1
?	HGMD curated	HGMD	CR052070	HGMD®	1

Disease gene annotation

gene	trait	source DB	source entry/link	
FCRL3	RHEUMATOID ARTHRITIS	OMIM	MIM:180300	OMIM®

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)
FCRL3	missense variant	ENST00000485028	?	ENSP00000434331	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000492769	?	ENSP00000435487	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000496769	?	ENSP00000473680	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000368186	?	ENSP00000357169	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000477837	?	ENSP00000433430	N/D	Aat/Gat	tolerated	benign	1
FCRL3	missense variant	ENST00000368184	NM_052939.3	ENSP00000357167	N/D	Aat/Gat	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FCRL3	ENST00000494724	ILMN_1691693	blood	1.12×10^{-20} (p-value)	MuTHER consortium	8
FCRL3	ENST00000485028					
FCRL3	ENST00000368186					
FCRL3	ENST00000477837					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000480682					
FCRL3	ENST00000494724	ILMN_1699599	blood	9.83×10^{-16} (p-value)	MuTHER consortium	8
FCRL3	ENST00000485028					
FCRL3	ENST00000477837					
FCRL3	ENST00000473231					
FCRL3	ENST00000468507					
FCRL3	ENST00000480682					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000473231	ILMN_1797428	blood	9.84×10^{-17} (p-value)	MuTHER consortium	8
FCRL3	ENST00000368184					
FCRL3	ENST00000468507					
FCRL3	?	ENSG00000160856	spleen	1.70×10^{-8} (p-value)	GTEx Portal V6	13
RP11-367J7.3	?	ENSG00000227217	spleen	9.09×10^{-8} (p-value)	GTEx Portal V6	5
FCRL3	?	ENSG00000160856	lung	2.74×10^{-9} (p-value)	GTEx Portal V6	13
RP11-367J7.3	?	ENSG00000227217	lung	5.02×10^{-8} (p-value)	GTEx Portal V6	11
RP11-367J7.3	?	ENSG00000227217	blood	7.86×10^{-13} (p-value)	GTEx Portal V6	13
FCRL3	?	ENSG00000160856	blood	2.00×10^{-19} (p-value)	GTEx Portal V6	13
FCRL1	?	ENSG00000163534	blood	8.70×10^{-7} (p-value)	GTEx Portal V6	4
FCRL3	?	ENSG00000160856	esophagus mucosa	5.44×10^{-9} (p-value)	GTEx Portal V6	9
ATP1A4	ENST00000470705	ILMN_1705208	b-cell	1.09×10^{-4} (p-value)	Fairfax et al.	3
ATP1A4	ENST00000466526					
FCRL3	ENST00000494724	ILMN_1691693	monocyte	1.27×10^{-6} (p-value)	Fairfax et al.	3
FCRL3	ENST00000485028		b-cell	7.43×10^{-33} (p-value)	Fairfax et al.	3
FCRL3	ENST00000477837					
FCRL3	ENST00000368186					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					
FCRL3	ENST00000480682					
FCRL3	ENST00000494724	ILMN_1699599	b-cell	2.37×10^{-30} (p-value)	Fairfax et al.	2
FCRL3	ENST00000485028					
FCRL3	ENST00000477837					
FCRL3	ENST00000473231					
FCRL3	ENST00000480682					
FCRL3	ENST00000468507					
FCRL3	ENST00000492769					
FCRL3	ENST00000368184					

FCRL3	ENST00000368184	ILMN_1797428	monocyte	1.02×10^{-4} (p-value)	Fairfax et al.	3
FCRL3	ENST00000473231		b-cell	5.93×10^{-26} (p-value)	Fairfax et al.	3
FCRL3	ENST00000468507					
FCRL3	?	ENSG00000160856	subcutaneous adipocytes	5.58×10^{-8} (p-value)	GTEx Portal V6	7

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
FCRL3	ENST00000457799	231093_at	chr1	blood	1.00×10^{-10} (p-value)	Dixon et al.	3
FCRL3	ENST00000368184						

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000671247 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	CTCF, H3K27me3
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF, H3K4me1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, H3K27me3
ENSR00000545969 (open chromatin region)	1	embryonic stem cell (H1ESC)	DNase1
		A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCRL3	upstream gene variant	283	ENST00000485028	?	ENSP00000434331	1
FCRL3	downstream gene variant, upstream gene variant	314	ENST00000478179	?	?	7
FCRL3	upstream gene variant	296	ENST00000494724	?	?	1
FCRL3	downstream gene variant, upstream gene variant	281	ENST00000496769	?	ENSP00000473680	2
FCRL3	upstream gene variant	587	ENST00000473231	?	?	6
FCRL3	upstream gene variant	2168	ENST00000468507	?	?	1
FCRL3	upstream gene variant	288	ENST00000368186	?	ENSP00000357169	1
FCRL3	upstream gene variant	296	ENST00000477837	?	ENSP00000433430	1
FCRL3	upstream gene variant	296	ENST00000480682	?	?	1
FCRL3	upstream gene variant	296	ENST00000492769	?	ENSP00000435487	1
FCRL3	upstream gene variant	169	ENST00000368184	NM_052939.3	ENSP00000357167	1
RP11-367J7.3	downstream gene variant	395	ENST00000453692	?	?	7
VDAC1P9	downstream gene variant	248	ENST00000412766	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCRL3	ENST00000477837	?	ENSP00000433430	5
FCRL3	ENST00000485028	?	ENSP00000434331	5
FCRL3	ENST00000480682	?	?	5
FCRL3	ENST00000368186	?	ENSP00000357169	5
FCRL3	ENST00000494724	?	?	5
FCRL3	ENST00000473231	?	?	2
FCRL3	ENST00000492769	?	ENSP00000435487	5
FCRL3	ENST00000368184	NM_052939.3	ENSP00000357167	5
FCRL3	ENST00000496769	?	ENSP00000473680	3
RP11-367J7.3	ENST00000453692	?	?	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCRL3	ENST00000477837	?	ENSP00000433430	1
FCRL3	ENST00000485028	?	ENSP00000434331	1
FCRL3	ENST00000368186	?	ENSP00000357169	1
FCRL3	ENST00000492769	?	ENSP00000435487	1
FCRL3	ENST00000368184	NM_052939.3	ENSP00000357167	1
FCRL3	ENST00000496769	?	ENSP00000473680	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCRL3	ENST00000480682	?	3
FCRL3	ENST00000494724	?	3
FCRL3	ENST00000473231	?	1