Locus 251 (rs11209026)

SNiPAcard

Block annotations

Block info

genomic range	chr1:67,684,934-67,743,552
block size	58,619 bp
variant count	26 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.033 [-2.084 – 2.716]	**gene(s) hit or close-by**	AL109843.1, C1orf141, IL23R, RNU4ATAC4P, RP11-131O15.2
phastCons	μ = 0.059 [0 – 0.959]	**eQTL gene(s)**	–
GERP++	μ = 0.161 [-4.62 – 5.19]	**potentially regulated gene(s)**	–
CADD score	μ = 3.253 [0.074 – 18.58]	**disease gene(s)**	IL23R

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link		variant(s)
Inflammatory bowel disease	$<8.00\times10^{-161}$	GWAS Catalog	23128233		1
Crohn's disease (time to surgery)	$<4.00\times10^{-14}$	GWAS Catalog	22936669		1
Crohn's disease (time to surgery)	$<1.00\times10^{-18}$	GWAS Catalog	22412388		1
Crohn's disease (time to surgery)	$<4.00\times10^{-21}$	GWAS Catalog	22293688		1
Ankylosing spondylitis	$<2.00\times10^{-17}$	GWAS Catalog	21743469		1
Ulcerative colitis	$<5.00\times10^{-28}$	GWAS Catalog	21297633		1
Crohn's disease (time to surgery)	$<1.00\times10^{-64}$	GWAS Catalog	21102463		1
Psoriasis	$<7.00\times10^{-7}$	GWAS Catalog	20953190		1
Crohn's disease (time to surgery)	$<1.00\times10^{-6}$	GWAS Catalog	20570966		1
Ankylosing spondylitis	$<9.00\times10^{-14}$	GWAS Catalog	20062062		1
Ulcerative colitis	$<3.00\times10^{-10}$	GWAS Catalog	19915572		1
Ulcerative colitis	$<1.00\times10^{-8}$	GWAS Catalog	19122664		1
Inflammatory bowel disease	$<7.00\times10^{-11}$	GWAS Catalog	18758464		1
Crohn's disease (time to surgery)	$<7.00\times10^{-63}$	GWAS Catalog	18587394		1
Crohn's disease (time to surgery)	$<1.00\times10^{-8}$	GWAS Catalog	17804789		3
Crohn's disease (time to surgery)	$<2.00\times10^{-18}$	GWAS Catalog	17447842		1
Inflammatory bowel diseases	4.59×10^{-11}	dbGaP	pha002847	dbGaP	2

Variant annotation

trait	type	source DB	source entry/link		Variant(s)
Psoriasis, protection against	protective	ClinVar	RCV000003255.2	ClinVar	1
Inflammatory bowel disease 17, protection against	protective	ClinVar	RCV000003254.2	ClinVar	1
?	HGMD curated	HGMD	CM066884	HGMD	1
INFLAMMATORY BOWEL DISEASE 17, PROTECTION AGAINST	OMIM curated	OMIM	MIM:607562	OMIM	1
INFLAMMATORY BOWEL DISEASE 17, PROTECTION AGAINST	UniProt/OMIM curated	UniProt	MIM:612261 (IL23R)	OMIM	1

Disease gene annotation

gene	trait	source DB	source entry/link
IL23R	PSORIASIS 7, SUSCEPTIBILITY TO	OMIM	MIM:605606
IL23R	INFLAMMATORY BOWEL DISEASE 17	OMIM	MIM:612261
IL23R	Behcet disease	OrphaNet	OrphaNet:117

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)
IL23R	missense variant	ENST00000425614	?	ENSP00000387640	Q/R	cAa/cGa	?	?	1
IL23R	missense variant	ENST00000347310	NM_144701.2	ENSP00000321345	Q/R	cAa/cGa	?	?	1

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000283180 (CTCF binding site)	1	embryonic stem cell (H1ESC)	CTCF, Rad21
		HSMMtube	H3K27me3
		blood (K562)	CTCF
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		blood (GM12878)	Rad21, CTCF
		A549	CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL109843.1	downstream gene variant, upstream gene variant	748	ENST00000408806	?	?	5
C1orf141	upstream gene variant	334	ENST00000371007	NM_001276351.1	ENSP00000360046	3
C1orf141	upstream gene variant	333	ENST00000448166	?	ENSP00000415519	3
IL23R	downstream gene variant	746	ENST00000473881	?	ENSP00000486667	1
IL23R	upstream gene variant, downstream gene variant	306	ENST00000395227	?	ENSP00000378652	2
IL23R	downstream gene variant	1173	ENST00000425614	?	ENSP00000387640	1
IL23R	downstream gene variant	442	ENST00000347310	NM_144701.2	ENSP00000321345	1
RNU4ATAC4P	downstream gene variant, upstream gene variant	772	ENST00000516307	?	?	4
RP11-131O15.2	upstream gene variant	183	ENST00000432257	?	?	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
C1orf141	ENST00000371007	NM_001276351.1	ENSP00000360046	3
C1orf141	ENST00000448166	?	ENSP00000415519	3
IL23R	ENST00000347310	NM_144701.2	ENSP00000321345	16
IL23R	ENST00000425614	?	ENSP00000387640	16

IL23R	ENST00000473881	?	ENSP00000486667	17
IL23R	ENST00000395227	?	ENSP00000378652	16

Locus 251 (sentinel SNP rs11209026 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.