Locus 16 (rs7551957)

SNiPAcard

Block annotations

Block info

genomic range	chr1:161,469,054-161,479,745
block size	10,692 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.167 [-0.902 – 0.932]	**gene(s) hit or close-by**	FCGR2A, RP11-25K21.6
phastCons	μ = 0.192 [0 – 0.836]	**eQTL gene(s)**	SDHC
GERP++	μ = -0.468 [-4.15 – 1.13]	**potentially regulated gene(s)**	FCGR2A
CADD score	μ = 2.723 [0.63 – 5.131]	**disease gene(s)**	SDHC

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
SYSTEMIC LUPUS ERYTHEMATOSUS	$<9.00\times10^{-6}$	GWAS Catalog	24871463	1
Inflammatory bowel disease	$<2.00\times10^{-38}$	GWAS Catalog	23128233	1
Kawasaki disease	$<7.00\times10^{-11}$	GWAS Catalog	22081228	1
Ulcerative colitis	$<2.00\times10^{-20}$	GWAS Catalog	21297633	1
Ulcerative colitis	$<2.00\times10^{-12}$	GWAS Catalog	19915573	1

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
Malaria, severe, susceptibility to	risk factor	ClinVar	RCV000054529.1	1
Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis	risk factor	ClinVar	RCV000015947.2	1
Lupus nephritis, susceptibility to	risk factor	ClinVar	RCV000015946.2	1
?	HGMD curated	HGMD	CM960640	1
LUPUS NEPHRITIS, SUSCEPTIBILITY TO	OMIM curated	OMIM	MIM:146790	1

Disease gene annotation

gene	trait	source DB	source entry/link
SDHC	PARAGANGLIOMA AND GASTRIC STROMAL SARCOMA	OMIM	MIM:606864
SDHC	PARAGANGLIOMAS 3	OMIM	MIM:605373
SDHC	COWDEN SYNDROME	OrphaNet	OrphaNet:201
SDHC	Gastrointestinal stromal tumor	OrphaNet	OrphaNet:44890
SDHC	Carney-Stratakis syndrome	OrphaNet	OrphaNet:97286
SDHC	Hereditary pheochromocytoma-paraganglioma	OrphaNet	OrphaNet:29072

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)
FCGR2A	missense variant	ENST00000367972	NM_021642.3	ENSP00000356949	H/R	cAt/cGt	tolerated	benign	1

FCGR2A	missense variant	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	H/R	cAt/cGt	tolerated	benign	1
FCGR2A	missense variant	ENST00000467525	?	ENSP00000476495	H/R	cAt/cGt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SDHC	ENST00000367975	ILMN_2323366	blood	1.21×10^{-4} (p-value)	Westra et al.	1
?	?	ILMN_1728639	blood	9.71×10^{-5} (p-value)	Westra et al.	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000374341	1	ENST00000367972, ENST00000271450, ENST00000471026, ENST00000467525	FCGR2A

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000286584 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K4me1
		HSMMtube	H3K27me3
		Osteobl	H3K27me3
		nervous (NH-A)	H3K27me3
		blood (K562)	H2AZ
		A549	H3K27me3
		blood (DND-41)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
FCGR2A	upstream gene variant	3422	ENST00000461298	?	?	3
FCGR2A	upstream gene variant	4715	ENST00000367972	NM_021642.3	ENSP00000356949	1
FCGR2A	upstream gene variant, downstream gene variant	2571	ENST00000497474	?	?	4
FCGR2A	upstream gene variant	4715	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	1
FCGR2A	upstream gene variant	3943	ENST00000459885	?	ENSP00000476411	3
FCGR2A	upstream gene variant, downstream gene variant	2652	ENST00000473080	?	?	4
FCGR2A	upstream gene variant	4742	ENST00000483665	?	ENSP00000440148	1
FCGR2A	upstream gene variant, downstream gene variant	56	ENST00000536731	?	ENSP00000441156	2
FCGR2A	downstream gene variant	2380	ENST00000482233	?	?	3
FCGR2A	upstream gene variant	4731	ENST00000467654	?	?	1
FCGR2A	upstream gene variant	4665	ENST00000486608	?	?	1
RP11-25K21.6	upstream gene variant	3221	ENST00000537821	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000367972	NM_021642.3	ENSP00000356949	2
FCGR2A	ENST00000271450	NM_001136219.1, NM_201563.5	ENSP00000271450	2
FCGR2A	ENST00000536731	?	ENSP00000441156	2
FCGR2A	ENST00000471026	?	?	2
FCGR2A	ENST00000467654	?	?	2
FCGR2A	ENST00000483665	?	ENSP00000440148	2
FCGR2A	ENST00000467525	?	ENSP00000476495	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
FCGR2A	ENST00000483665	?	ENSP00000440148	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
FCGR2A	ENST00000471026	?	1
FCGR2A	ENST00000467654	?	1