Locus 15 (rs3776081)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,519,260-149,537,194
block size	17,935 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.217 [-0.624 – 0.211]	gene(s) hit or close-by	PDGFRB
phastCons	μ = 0.001 [0 – 0.002]	eQTL gene(s)	CDX1, CSF1R, PDGFRB
GERP++	μ = 0.540 [-0.121 – 1.03]	potentially regulated gene(s)	–
CADD score	μ = 6.404 [4.963 – 8.056]	disease gene(s)	PDGFRB, CSF1R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
PDGFRB	Bilateral striopallidodentate calcinosis	OrphaNet	OrphaNet:1980
PDGFRB	Unclassified chronic myeloproliferative disease	OrphaNet	OrphaNet:86830
PDGFRB	Chronic myelomonocytic leukemia	OrphaNet	OrphaNet:98823
PDGFRB	Idiopathic hypereosinophilic syndrome	OrphaNet	OrphaNet:3260
PDGFRB	Infantile myofibromatosis 1	OrphaNet	OrphaNet:2591
PDGFRB	Myeloid neoplasm associated with PDGFRB rearrangement	OrphaNet	OrphaNet:168950
CSF1R	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	OrphaNet	OrphaNet:313808

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDGFRB	ENST00000520579	ILMN_1815057	blood	8.08×10^{-39} (p-value)	MuTHER consortium	1
PDGFRB	ENST00000261799					
CSF1R	ENST00000509861	ILMN_1686623	blood	5.51×10^{-7} (p-value)	Westra et al.	1
CSF1R	ENST00000504875					
CSF1R	ENST00000286301					
CDX1	?	ENSG00000113722	tibial nerve	5.45×10^{-13} (p-value)	GTEx Portal V6	2
PDGFRB	?	ENSG00000113721	EBV lymphocytes	6.13×10^{-8} (p-value)	GTEx Portal V6	2
PDGFRB	ENST00000520579	ILMN_1815057	b-cell	5.54×10^{-6} (p-value)	Fairfax et al.	1
PDGFRB	ENST00000261799					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001293753 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K4me1, USF1
		HSMMtube	H3K4me1, H3K9ac, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K27me3, DNase1, Egr1, PU1, Cjun
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, H3K4me1, H3K36me3
		muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	FOSL2, Jund, H3K4me3, PolII, TAF1, ELF1, USF1, H3K4me1, p300, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, DNase1
		lung (IMR90)	H3K56ac, H3K79me2, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H3K9ac, H3K4me1, H2BK12ac, DNase1, H4K8ac, H4K91ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me1, H3K4me2, H3K36me3, H3K27ac
		skin (NHEK)	H3K27me3, DNase1
		NHLF	H3K27ac, H3K36me3, H3K9ac, H3K4me1, DNase1
		Osteobl	H3K36me3, H3K4me1, H3K4me2, H3K27ac
		blood (DND-41)	H3K27me3
		breast (HMEC)	H3K27ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Cjun, Jund, H3K4me2, H3K27ac, H3K4me1, Max
		monocytes (Monocytes-CD14+)	H3K27ac, DNase1
		endothelium (HUVEC)	H3K27me3
		A549	H3K4me3, H3K4me2, DNase1, H3K27ac
ENSR00001698907 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K27me3, Rad21, CTCF
		HSMMtube	DNase1, CTCF
		blood (K562)	H3K27me3, PolII, Egr1, Rad21, H2AZ, H3K4me2, CTCF, DNase1
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, CTCF, H3K4me1
		muscle (HSMM)	H2AZ, CTCF
		liver (HepG2)	Rad21, H2AZ, H3K4me2, CTCF, H3K27me3, DNase1, H3K4me3
		blood (GM12878)	DNase1, EBF1, H2AZ, Rad21, CTCF
		lung (IMR90)	DNase1, CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	H3K27me3, CTCF
		NHLF	DNase1, H3K4me1, CTCF, H3K27ac
		Osteobl	CTCF, H3K27ac
		blood (DND-41)	CTCF, H3K27ac, H3K27me3, H3K4me1
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K4me1, CTCF, DNase1
		monocytes (Monocytes-CD14+)	H3K27me3, CTCF
		endothelium (HUVEC)	H3K36me3, DNase1, H3K27me3, CTCF
		A549	H3K4me3, H3K4me2, CTCF, DNase1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	2260	ENST00000517957	?	ENSP00000430715	1
PDGFRB	upstream gene variant	1771	ENST00000517660	?	?	1
PDGFRB	downstream gene variant, upstream gene variant	867	ENST00000523456	?	?	2
PDGFRB	upstream gene variant	1772	ENST00000520579	?	ENSP00000430026	1
PDGFRB	upstream gene variant	2004	ENST00000517488	?	ENSP00000429218	1
PDGFRB	upstream gene variant	1771	ENST00000261799	NM_002609.3	ENSP00000261799	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000517660	?	?	2
PDGFRB	ENST00000517488	?	ENSP00000429218	2

PDGFRB	ENST00000520579	?	ENSP00000430026	2
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	2

Locus 15 (sentinel SNP rs3776081 + 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.