SNiPAcard

Block annotations

Block info

genomic range	chr5:149,504,115-149,505,624
block size	1,510 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.645 [-2.666 – -0.656]	**gene(s) hit or close-by**	PDGFRB
phastCons	μ = 0.000 [0 – 0]	**eQTL gene(s)**	CSF1R, PDGFRB
GERP++	μ = -2.577 [-3.26 – -1.88]	**potentially regulated gene(s)**	–
CADD score	μ = 4.338 [2.553 – 6.987]	**disease gene(s)**	PDGFRB, CSF1R

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PDGFRB	JUVENILE MYELOMONOCYTIC LEUKEMIA	OMIM	MIM:607785
PDGFRB	BASAL GANGLIA CALCIFICATION, IDIOPATHIC, 4	OMIM	MIM:615007
PDGFRB	LEUKEMIA, ACUTE MYELOID	OMIM	MIM:601626
PDGFRB	MYOFIBROMATOSIS, INFANTILE, 1	OMIM	MIM:228550
PDGFRB	MYELOPROLIFERATIVE DISORDER, CHRONIC, WITH EOSINOPHILIA	OMIM	MIM:131440
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550
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	2.97×10^{-7} (p-value)	MuTHER consortium	2
PDGFRB	ENST00000261799					
CSF1R	ENST00000509861	ILMN_1686623	blood	3.96×10^{-5} (p-value)	Westra et al.	1
CSF1R	ENST00000504875					
CSF1R	ENST00000286301					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	2315	ENST00000521723	?	?	3

PDGFRB	upstream gene variant	2350	ENST00000519575	?	?	3
PDGFRB	upstream gene variant	223	ENST00000520229	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000520579	?	ENSP00000430026	3
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	3

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PDGFRB	ENST00000520229	?	1