Locus 329 (rs11746309)

SNiPAcard

Block annotations

Block info

genomic range	chr5:149,472,918-149,481,535
block size	8,618 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.294 [0.136 – 0.621]	**gene(s) hit or close-by**	CSF1R, RPL7P1, Y_RNA
phastCons	μ = 0.017 [0.002 – 0.042]	**eQTL gene(s)**	CSF1R, PDGFRB
GERP++	μ = 0.287 [-0.693 – 1.97]	**potentially regulated gene(s)**	–
CADD score	μ = 2.375 [1.134 – 3.976]	**disease gene(s)**	CSF1R, PDGFRB

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
CSF1R	LEUKOENCEPHALOPATHY, DIFFUSE HEREDITARY, WITH SPHEROIDS	OMIM	MIM:221820
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	Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia	OrphaNet	OrphaNet:313808
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
PDGFRB	Familial Infantile Myofibromatosis	DECIPHER	MIM:228550

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PDGFRB	ENST00000520579	ILMN_1815057	blood	1.19×10^{-11} (p-value)	MuTHER consortium	4
PDGFRB	ENST00000261799					
CSF1R	?	ENSG00000182578	stomach	4.89×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
RPL7P1	upstream gene variant, downstream gene variant	947	ENST00000468161	?	?	2
Y_RNA	downstream gene variant	589	ENST00000362539	?	?	5

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CSF1R	ENST00000286301	NM_005211.3, NM_001288705.1	ENSP00000286301	6
CSF1R	ENST00000511344	?	ENSP00000421174	6