SNiPAcard

Block annotations

Block info

genomic range	chr5:149,521,238-149,521,238
block size	1 bp
variant count	1 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	-0.925	**gene(s) hit or close-by**	PDGFRB
phastCons	0	**eQTL gene(s)**	–
GERP++	-0.18	**potentially regulated gene(s)**	–
CADD score	7.735	**disease gene(s)**	PDGFRB

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
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

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

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
PDGFRB	upstream gene variant	4238	ENST00000517957	?	ENSP00000430715	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PDGFRB	ENST00000520579	?	ENSP00000430026	1
PDGFRB	ENST00000517488	?	ENSP00000429218	1
PDGFRB	ENST00000517660	?	?	1
PDGFRB	ENST00000261799	NM_002609.3	ENSP00000261799	1