SNiPAcard

Block annotations

Block info

genomic range	chr11:120,098,044-120,100,867
block size	2,824 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.208 [-2.697 – 2.393]	**gene(s) hit or close-by**	OAF
phastCons	μ = 0.129 [0 – 1]	**eQTL gene(s)**	POU2F3
GERP++	μ = -0.794 [-5.98 – 5.26]	**potentially regulated gene(s)**	ARHGEF12, CTD-2523D13.2, OAF, POU2F3, PVRL1, TMEM136, TRIM29
CADD score	μ = 4.439 [0.537 – 15.33]	**disease gene(s)**	PVRL1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
PVRL1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME	OMIM	MIM:225060
PVRL1	CLEFT LIP/PALATE-ECTODERMAL DYSPLASIA SYNDROME	DECIPHER	MIM:225060
PVRL1	Zlotogora-Ogur syndrome	OrphaNet	OrphaNet:3253

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)
OAF	missense variant	ENST00000328965	NM_178507.2	ENSP00000332613	H/R	cAc/cGc	tolerated	benign	1
OAF	missense variant	ENST00000531220	?	ENSP00000431865	H/R	cAc/cGc	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
POU2F3	?	ENSG00000137709	subcutaneous adipocytes	6.18×10^{-8} (p-value)	GTEx Portal V6	8

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000047292	1	ENST00000328965, ENST00000531220	OAF

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)

ENCE00000125097	1	ENCP00000015007	CTD-2523D13.2 PVRL1
		ENCP00000015012	TRIM29
		ENCP00000015027	OAF
		ENCP00000015028	POU2F3
		ENCP00000015032	TMEM136
		ENCP00000015034	ARHGEF12

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000112413 (CTCF binding site)	3	embryonic stem cell (H1ESC)	H3K27me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, H3K27me3
		blood (K562)	H3K27me3, Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, H3K36me3, PolII, Rad21, CTCF
		blood (GM12878)	CTCF, Rad21
		lung (IMR90)	CTCF, H3K36me3
		nervous (NH-A)	CTCF
		skin (NHEK)	H3K36me3, CTCF
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K27me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
OAF	downstream gene variant	571	ENST00000531220	?	ENSP00000431865	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
OAF	ENST00000328965	NM_178507.2	ENSP00000332613	4
OAF	ENST00000531220	?	ENSP00000431865	4

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
OAF	ENST00000328965	NM_178507.2	ENSP00000332613	3