

Block annotations

Block info

genomic range	chr6:123,121,472-123,128,536 <i>e!</i>
block size	7,065 bp
variant count	4 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = 1.207$ [-1.366 – 6.514]	gene(s) hit or close-by	SMPDL3A <i>e!</i>
phastCons	$\mu = 0.250$ [0 – 1]	eQTL gene(s)	SMPDL3A <i>e!</i>
GERP++	$\mu = 0.172$ [-3.94 – 5.72]	potentially regulated gene(s)	SMPDL3A <i>e!</i>
CADD score	$\mu = 5.927$ [0.363 – 14.62]	disease gene(s)	–

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)
SMPDL3A <i>e!</i>	missense variant	ENST00000539041 <i>e!</i>	NM_001286138.1	ENSP00000442152	S/P	Tct/Cct	?	?	1
SMPDL3A <i>e!</i>	missense variant	ENST00000368440 <i>e!</i>	NM_006714.4	ENSP00000357425	S/P	Tct/Cct	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
SMPDL3A <i>e!</i>	?	ENSG00000172594 <i>e!</i>	tibial nerve	2.43×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1
SMPDL3A <i>e!</i>	?	ENSG00000172594 <i>e!</i>	transformed fibroblasts	1.28×10 <sup>-5</sup> (p-value)	GTEX Portal V6 <i>e!</i>	1

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000715093 <i>e!</i>	1	ENST00000368440 <i>e!</i> , ENST00000539041 <i>e!</i>	SMPDL3A <i>e!</i>

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001226031 <i>e!</i> (promoter flanking region)	2	NHLF Osteobl blood (DND-41) muscle (HSMM) cervix (HeLa-S3) monocytes (Monocytes-CD14+) liver (HepG2) lung (IMR90) blood (GM12878) nervous (NH-A) skin (NHEK)	DNase1 H3K4me2 H3K27me3 DNase1, H3K36me3, H3K4me2 DNase1 H3K36me3 DNase1, H3K4me1, H3K4me2, FOXA1, H3K36me3 DNase1, H3K36me3 BATF DNase1 H3K4me1, H3K4me2, DNase1

ENSR00001226034 <i>e!</i> (CTCF binding site)	1	monocytes (Monocytes-CD14+) lung (IMR90)	H3K36me3 H3K27ac
--	---	---	---------------------

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
SMPDL3A <i>e!</i>	downstream gene variant	2982	ENST00000487215 <i>e!</i>	?	?	2

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
SMPDL3A <i>e!</i>	ENST00000539041 <i>e!</i>	NM_001286138.1	ENSP00000442152 <i>e!</i>	3
SMPDL3A <i>e!</i>	ENST00000368440 <i>e!</i>	NM_006714.4	ENSP00000357425 <i>e!</i>	3

