SNiPAcard

Block annotations

Block info

genomic range	chr6:160,476,945-160,513,793
block size	36,849 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.604 [-2.574 – 0.77]	**gene(s) hit or close-by**	IGF2R, RP11-288H12.3
phastCons	μ = 0.001 [0 – 0.006]	**eQTL gene(s)**	IGF2R, RP11-288H12.3
GERP++	μ = -1.707 [-6.01 – 1.86]	**potentially regulated gene(s)**	LPA, RP1-276N6.2
CADD score	μ = 2.880 [0.501 – 6.396]	**disease gene(s)**	–

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
isobutyrylcarnitine	5.24×10^{-9}	Metabolomics GWAS Server	24816252	8

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RP11-288H12.3	?	ENSG00000213073	ovary	2.23×10^{-6} (p-value)	GTEx Portal V6	1
RP11-288H12.3	?	ENSG00000213073	blood	2.66×10^{-14} (p-value)	GTEx Portal V6	9
RP11-288H12.3	?	ENSG00000213073	transformed fibroblasts	4.66×10^{-8} (p-value)	GTEx Portal V6	8
IGF2R	?	ENSG00000197081	aorta	4.15×10^{-6} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	thyroid	1.78×10^{-6} (p-value)	GTEx Portal V6	8
RP11-288H12.3	?	ENSG00000213073	EBV lymphocytes	6.38×10^{-7} (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000468394	1	ENCP00000050541	RP1-276N6.2
		ENCP00000050544	LPA

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001232516 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		blood (K562)	H3K36me3
		skin (NHEK)	H3K36me3
		breast (HMEC)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
IGF2R	downstream gene variant	2956	ENST00000487607	?	?	1
IGF2R	upstream gene variant	3686	ENST00000475584	?	?	1
IGF2R	upstream gene variant, downstream gene variant	2199	ENST00000475834	?	ENSP00000473880	3
RP11-288H12.3	upstream gene variant	662	ENST00000366273	?	?	2
RP11-288H12.3	upstream gene variant	321	ENST00000569097	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000356956	NM_000876.2	ENSP00000349437	9
IGF2R	ENST00000487607	?	?	1
IGF2R	ENST00000475834	?	ENSP00000473880	3

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
IGF2R	ENST00000475834	?	ENSP00000473880	1