Locus 347 (rs939421)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,576,081-46,585,153
block size	9,073 bp
variant count	3 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.659 [-3.044 – -0.916]	**gene(s) hit or close-by**	LRRC2
phastCons	μ = 0.003 [0 – 0.009]	**eQTL gene(s)**	LRRC2, LRRC2-AS1, TDGF1
GERP++	μ = -1.157 [-2.2 – 0.158]	**potentially regulated gene(s)**	–
CADD score	μ = 1.710 [0.009 – 2.758]	**disease gene(s)**	TDGF1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
TDGF1	Semilobar holoprosencephaly	OrphaNet	OrphaNet:220386
TDGF1	Alobar holoprosencephaly	OrphaNet	OrphaNet:93925
TDGF1	Microform holoprosencephaly	OrphaNet	OrphaNet:280200
TDGF1	Midline interhemispheric variant of holoprosencephaly	OrphaNet	OrphaNet:93926
TDGF1	Septopreoptic holoprosencephaly	OrphaNet	OrphaNet:280195
TDGF1	Lobar holoprosencephaly	OrphaNet	OrphaNet:93924

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TDGF1	?	ENSG00000241186	skeletal muscle	2.49×10^{-7} (p-value)	GTEx Portal V6	3
LRRC2-AS1	?	ENSG00000268324	skeletal muscle	1.91×10^{-10} (p-value)	GTEx Portal V6	3
LRRC2	?	ENSG00000163827	testis	3.46×10^{-9} (p-value)	GTEx Portal V6	3
LRRC2-AS1	?	ENSG00000268324	testis	7.24×10^{-12} (p-value)	GTEx Portal V6	3
TDGF1	?	ENSG00000241186	tibial nerve	2.97×10^{-5} (p-value)	GTEx Portal V6	2
LRRC2	?	ENSG00000163827	transformed fibroblasts	1.11×10^{-11} (p-value)	GTEx Portal V6	3
TDGF1	?	ENSG00000241186	left ventricle	9.43×10^{-6} (p-value)	GTEx Portal V6	2

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001362985 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, H3K36me3
		Osteobl	H3K4me2, H3K27ac, H2AZ
		blood (K562)	H3K27me3

blood (DND-41)	H3K4me1, H3K27ac, H3K9ac
skin (NHDF-AD)	DNase1, H3K4me2
muscle (HSMM)	H3K36me3
monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K4me3
endothelium (HUVEC)	H3K27me3
liver (HepG2)	DNase1
blood (GM12878)	PU1, MEF2C, BATF, MEF2A, H3K4me1, Tcf12, DNase1, IRF4, BCL11A, Pax5, H3K27ac, H3K4me2
lung (IMR90)	DNase1, H3K36me3
A549	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LRRC2	downstream gene variant	1488	ENST00000496388	?	?	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC2	ENST00000296144	?	ENSP00000296144	3
LRRC2	ENST00000395905	NM_024512.4	ENSP00000379241	3