Locus 9 (rs3806701)

SNiPAcard

Block annotations

Block info

genomic range	chr3:46,596,264-46,631,058
block size	34,795 bp
variant count	9 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.261 [-1.057 – 0.212]	gene(s) hit or close-by	AC104304.1, LRRC2, LRRC2-AS1, TDGF1
phastCons	μ = 0.047 [0 – 0.328]	eQTL gene(s)	ALS2CL, LRRC2, LRRC2-AS1, TDGF1
GERP++	μ = -0.249 [-4.4 – 1.74]	potentially regulated gene(s)	AC104304.1, CCR2, LRRC2, LTF, PRSS44, PRSS50
CADD score	μ = 4.291 [1.295 – 8.84]	disease gene(s)	TDGF1, LTF, CCR2

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
LTF	SPECIFIC GRANULE DEFICIENCY	OMIM	MIM:245480
CCR2	HUMAN IMMUNODEFICIENCY VIRUS TYPE 1, SUSCEPTIBILITY TO	OMIM	MIM:609423
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	atrial appendage	6.64×10^{-21} (p-value)	GTEx Portal V6	9
ALS2CL	?	ENSG00000178038	transformed fibroblasts	9.66×10^{-7} (p-value)	GTEx Portal V6	5
LRRC2	?	ENSG00000163827	transformed fibroblasts	7.45×10^{-33} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	breast	1.41×10^{-27} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	skeletal muscle	1.27×10^{-57} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	putamen	2.37×10^{-10} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	unexposed skin	1.15×10^{-20} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	sun exposed skin	1.16×10^{-19} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	ovary	5.21×10^{-18} (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	ovary	2.35×10^{-6} (p-value)	GTEx Portal V6	1
TDGF1	?	ENSG00000241186	left ventricle	2.50×10^{-37} (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	subcutaneous adipocytes	6.28×10^{-55} (p-value)	GTEx Portal V6	9

TDGF1	?	ENSG00000241186	caudate basal ganglia	2.40×10-12 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	visceral adipocytes	2.25×10-32 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cortex	5.06×10-15 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	tibial nerve	8.12×10-48 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	pituitary	2.18×10-9 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	anterior cingulate cortex	1.80×10-10 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	esophagus mucosa	2.71×10-27 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	testis	4.56×10-17 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	testis	1.17×10-7 (p-value)	GTEx Portal V6	9
LRRC2-AS1	?	ENSG00000268324	testis	1.42×10-5 (p-value)	GTEx Portal V6	4
TDGF1	?	ENSG00000241186	hypothalamus	1.44×10-10 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	frontal cortex	7.78×10-15 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	uterus	3.21×10-13 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	muscularis mucosae	1.23×10-40 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	lung	1.26×10-27 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	tibial artery	1.66×10-8 (p-value)	GTEx Portal V6	7
TDGF1	?	ENSG00000241186	prostate	2.11×10-13 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	transverse colon	3.56×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	terminal ileum	2.23×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	nucleus accumbens	1.76×10-12 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	spleen	2.86×10-21 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	coronary artery	1.44×10-14 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	sigmoid colon	1.13×10-25 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	vagina	2.24×10-9 (p-value)	GTEx Portal V6	7
TDGF1	?	ENSG00000241186	gastroesophageal junction	2.89×10-25 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	adrenal gland	1.93×10-27 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	adrenal gland	8.32×10-27 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cerebellar hemisphere	5.18×10-8 (p-value)	GTEx Portal V6	9
TDGF1	?	ENSG00000241186	cerebellum	8.08×10-9 (p-value)	GTEx Portal V6	9
LRRC2	?	ENSG00000163827	cerebellum	9.29×10-7 (p-value)	GTEx Portal V6	5
TDGF1	?	ENSG00000241186	hippocampus	2.99×10-9 (p-value)	GTEx Portal V6	9

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337772	1	ENCP00000036261	LRRC2
		ENCP00000036273	PRSS50
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036258	LTF

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001363001 (TF binding site)	3	embryonic stem cell (H1ESC)	Rad21, TAF7, H3K27ac, HDAC2, H3K4me2, SP1, H3K4me3, DNase1, PolII, H3K27me3, H3K36me3
		endothelium (HUVEC)	H3K27me3
		blood (K562)	H3K27me3
ENSR00001363008 (TF binding site)	2	embryonic stem cell (H1ESC)	DNase1, H3K36me3, PolII
		lung (IMR90)	H3K27me3
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC104304.1	upstream gene variant	4217	ENST00000583198	?	?	1
LRRC2-AS1	upstream gene variant	2624	ENST00000599511	?	?	1
LRRC2-AS1	upstream gene variant	2630	ENST00000614743	?	?	1
TDGF1	upstream gene variant	1394	ENST00000459867	?	?	4
TDGF1	upstream gene variant	3402	ENST00000505797	?	?	2
TDGF1	upstream gene variant	1519	ENST00000493282	?	?	3
TDGF1	upstream gene variant	17	ENST00000471721	?	?	4
TDGF1	upstream gene variant, downstream gene variant	1008	ENST00000296145	NM_003212.3	ENSP00000296145	5
TDGF1	downstream gene variant	4493	ENST00000542931	NM_001174136.1	ENSP00000446375	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LRRC2	ENST00000296144	?	ENSP00000296144	5
LRRC2	ENST00000496388	?	?	1
LRRC2	ENST00000395905	NM_024512.4	ENSP00000379241	1
LRRC2	ENST00000469912	?	?	1
TDGF1	ENST00000542931	NM_001174136.1	ENSP00000446375	4
TDGF1	ENST00000505797	?	?	4

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
TDGF1	ENST00000296145	NM_003212.3	ENSP00000296145	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)

TDGF1	ENST00000493282	?	1

Locus 9 (sentinel SNP rs3806701 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.