SNiPAcard

Block annotations

Block info

genomic range	chr3:46,641,719-46,647,381
block size	5,663 bp
variant count	20 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.535 [-2.28 – 0.735]	**gene(s) hit or close-by**	TDGF1
phastCons	μ = 0.164 [0 – 0.712]	**eQTL gene(s)**	ATRIP, PRSS46, TDGF1, TREX1, TREX1
GERP++	μ = -0.251 [-1.53 – 0.559]	**potentially regulated gene(s)**	AC104304.1, CCR2, CCR3, LRRC2, PRSS44, PRSS50
CADD score	μ = 4.290 [0.163 – 14.08]	**disease gene(s)**	TDGF1, TREX1, TREX1, CCR2, ATRIP

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
TDGF1	TERATOCARCINOMA-DERIVED GROWTH FACTOR 1	OMIM	MIM:187395
TREX1	VASCULOPATHY, RETINAL, WITH CEREBRAL LEUKODYSTROPHY	OMIM	MIM:192315
TREX1	SYSTEMIC LUPUS ERYTHEMATOSUS	OMIM	MIM:152700
TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
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TREX1	CHILBLAIN LUPUS 1	OMIM	MIM:610448
TREX1	AICARDI-GOUTIERES SYNDROME 1	OMIM	MIM:225750
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
ATRIP	Seckel syndrome	OrphaNet	OrphaNet:808
TREX1	Aicardi-Goutieres Syndrome	OrphaNet	OrphaNet:51
TREX1	HERNS syndrome	OrphaNet	OrphaNet:63261
TREX1	CHILBLAIN LUPUS	OrphaNet	OrphaNet:90280
TREX1	Cerebroretinal vasculopathy	OrphaNet	OrphaNet:3421
TREX1	Hereditary vascular retinopathy	OrphaNet	OrphaNet:71291
TREX1	Aicardi-Goutieres syndrome 1, dominant and recessive	DECIPHER	MIM:225750
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Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PRSS46	?	ENSG00000261603	muscularis mucosae	1.66×10-6 (p-value)	GTEx Portal V6	19
TDGF1	?	ENSG00000241186	skeletal muscle	1.34×10-6 (p-value)	GTEx Portal V6	16

TREX1	ENST00000436480	ILMN_2370464	b-cell	4.74×10-4 (p-value)	Fairfax et al.	1
ATRIP	ENST00000320211					
ATRIP	ENST00000346691					
TREX1	ENST00000395677					
TREX1	ENST00000296443					
TREX1	ENST00000424906					
ATRIP	ENST00000412052					
TREX1	ENST00000433541					
TREX1	ENST00000629913					
TREX1	ENST00000357105					
TREX1	ENST00000625293					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000337725	1	ENCP00000036261	LRRC2
		ENCP00000036248	CCR2
		ENCP00000036263	AC104304.1
		ENCP00000036262	AC104304.1
		ENCP00000036277	PRSS44
		ENCP00000036244	CCR3
		ENCP00000036274	PRSS50
		ENCP00000036273	PRSS50

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001674640 (enhancer)	4	HSMMtube	H3K27me3
		blood (K562)	H3K27me3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
TDGF1	ENST00000505797	?	?	20