Locus 119 (rs556990)

SNiPAcard

Block annotations

Block info

genomic range	chr13:113,818,708-113,910,611
block size	91,904 bp
variant count	79 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.775 [-6.801 – 1.266]	gene(s) hit or close-by	CUL4A, PCID2, PROZ, RP11-98F14.11
phastCons	μ = 0.029 [0 – 0.987]	eQTL gene(s)	ADAMDEC1, CUL4A, F10, PCID2, PROZ, RP11-98F14.11
GERP++	μ = -1.100 [-8.08 – 2.42]	potentially regulated gene(s)	ADPRHL1, ATP11A, ATP4B, CUL4A, DCUN1D2, F7, GRTP1, MCF2L, PCID2, PROZ, TFDP1, TMCO3
CADD score	μ = 3.026 [0.009 – 15.28]	disease gene(s)	PROZ, ATP11A, F10, F7

Trait annotations

Variant annotation

trait	type	source DB	source entry/link	Variant(s)
?	HGMD curated	HGMD	CS040554	1

Disease gene annotation

gene	trait	source DB	source entry/link	
PROZ	Cerebral sinovenous thrombosis	OrphaNet	OrphaNet:329217	orphanet
ATP11A	Idiopathic pulmonary fibrosis	OrphaNet	OrphaNet:2032	orphanet
F10	Congenital factor X deficiency	OrphaNet	OrphaNet:328	orphanet
F7	Congenital factor VII deficiency	OrphaNet	OrphaNet:327	orphanet
F10	FACTOR X DEFICIENCY	OMIM	MIM:227600	OMIM
F7	FACTOR VII DEFICIENCY	OMIM	MIM:227500	OMIM

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
PROZ	?	ENSG00000126231	pancreas	1.30×10^{-16} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	muscularis mucosae	1.65×10^{-7} (p-value)	GTEx Portal V6	69
RP11-98F14.11	?	ENSG00000269125	muscularis mucosae	6.60×10^{-6} (p-value)	GTEx Portal V6	1
PROZ	?	ENSG00000126231	stomach	8.50×10^{-12} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	lung	1.04×10^{-15} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	tibial nerve	1.90×10^{-12} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	transformed fibroblasts	2.61×10^{-7} (p-value)	GTEx Portal V6	64
PROZ	?	ENSG00000126231	blood	4.04×10^{-14} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	tibial artery	1.42×10^{-14} (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	thyroid	1.85×10^{-16} (p-value)	GTEx Portal V6	78

RP11-98F14.11	?	ENSG00000269125	thyroid	1.43×10-8 (p-value)	GTEx Portal V6	51
PROZ	?	ENSG00000126231	cerebellar hemisphere	1.80×10-6 (p-value)	GTEx Portal V6	3
PROZ	?	ENSG00000126231	adrenal gland	8.50×10-8 (p-value)	GTEx Portal V6	33
PCID2	?	ENSG00000126226	testis	7.95×10-12 (p-value)	GTEx Portal V6	77
PROZ	?	ENSG00000126231	testis	2.75×10-13 (p-value)	GTEx Portal V6	75
RP11-98F14.11	?	ENSG00000269125	testis	3.71×10-7 (p-value)	GTEx Portal V6	19
PROZ	?	ENSG00000126231	cerebellum	8.07×10-9 (p-value)	GTEx Portal V6	78
RP11-98F14.11	?	ENSG00000269125	cerebellum	2.38×10-6 (p-value)	GTEx Portal V6	12
PROZ	?	ENSG00000126231	transverse colon	1.99×10-11 (p-value)	GTEx Portal V6	78
PROZ	?	ENSG00000126231	subcutaneous adipocytes	2.56×10-8 (p-value)	GTEx Portal V6	51
PCID2	ENST00000375477	ILMN_1788024	blood	1.85×10-7 (p-value)	MuTHER consortium	30
PCID2	ENST00000375459					
PCID2	ENST00000462653					
PCID2	ENST00000463102					
PCID2	ENST00000473462					
PCID2	ENST00000493650					
PCID2	ENST00000375479					
PCID2	ENST00000622406					
PCID2	ENST00000337344					
PCID2	ENST00000246505					
PCID2	ENST00000375457					
PROZ	?	ENSG00000126231	spleen	1.52×10-9 (p-value)	GTEx Portal V6	47
PROZ	?	ENSG00000126231	esophagus mucosa	4.90×10-8 (p-value)	GTEx Portal V6	76
PROZ	?	ENSG00000126231	atrial appendage	7.13×10-7 (p-value)	GTEx Portal V6	40
PROZ	?	ENSG00000126231	breast	2.74×10-6 (p-value)	GTEx Portal V6	13
PROZ	?	ENSG00000126231	prostate	1.94×10-7 (p-value)	GTEx Portal V6	15
PCID2	ENST00000375477	ILMN_1788024	blood	1.36×10-5 (p-value)	Westra et al.	1
PCID2	ENST00000375459					
PCID2	ENST00000462653					
PCID2	ENST00000463102					
PCID2	ENST00000473462					
PCID2	ENST00000493650					
PCID2	ENST00000375479					
PCID2	ENST00000622406					
PCID2	ENST00000337344					
PCID2	ENST00000246505					
PCID2	ENST00000375457					
F10	?	ENSG00000126218	left ventricle	1.10×10-7 (p-value)	GTEx Portal V6	27
PROZ	?	ENSG00000126231	sigmoid colon	2.45×10-6 (p-value)	GTEx Portal V6	10
CUL4A	?	ENSG00000139842	skeletal muscle	5.73×10-6 (p-value)	GTEx Portal V6	11

trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
ADAMDEC1	?	ENSG00000134028	chr8	blood	1.07×10^{-2} (q-value)	SeeQTL DB (HapMap)	3

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000019252		
ENCP00000019255		

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000165802	1	ENCP00000019270	ADPRHL1
ENCE00000165363	1	ENCP00000019222	ATP11A
		ENCP00000019281	TMCO3
		ENCP00000019218	ATP11A
ENCE00000165706	1	ENCP00000019286	ATP4B
		ENCP00000019258	CUL4A PCID2
ENCE00000165538	1	ENCP00000019231	MCF2L
ENCE00000165742	1	ENCP00000019261	CUL4A
		ENCP00000019279	TMCO3
		ENCP00000019278	TMCO3
ENCE00000165366	1	ENCP00000019222	ATP11A
		ENCP00000019281	TMCO3
		ENCP00000019218	ATP11A
ENCE00000165845	1	ENCP00000019276	DCUN1D2
ENCE00000165690	1	ENCP00000019252	PROZ
ENCE00000165334	1	ENCP00000019241	MCF2L
		ENCP00000019243	MCF2L
		ENCP00000019284	TFDP1
		ENCP00000019230	MCF2L
		ENCP00000019268	GRTP1
		ENCP00000019220	ATP11A
		ENCP00000019286	ATP4B
		ENCP00000019224	MCF2L
		ENCP00000019274	DCUN1D2
		ENCP00000019233	MCF2L
		ENCP00000019223	ATP11A
		ENCP00000019245	F7
		ENCP00000019259	PCID2
		ENCP00000019217	ATP11A
		ENCP00000019251	PROZ

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000520923 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000520925 (TF binding site)	1	embryonic stem cell (H1ESC)	H3K36me3
		endothelium (HUVEC)	DNase1, Cjun
		lung (IMR90)	H3K36me3
ENSR00000520926	1	NHLF	H3K36me3

(enhancer)		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000520927 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H4K8ac, H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001036786 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K36me3, DNase1, H3K27ac
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K36me3
		lung (IMR90)	H4K91ac, H3K56ac, H2BK15ac, H3K23ac, H2BK12ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2AK5ac, H4K8ac, DNase1, H3K9ac
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	H3K36me3
		NHLF	H3K27ac, H3K9ac, H3K4me1, DNase1, H3K36me3
		Osteobl	H3K4me2, H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3, H3K27ac
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00001512527 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K36me3, DNase1
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000520931 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		blood (K562)	H3K36me3, Cjun, Junb, FOSL1
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	H3K79me2, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	FOSL2, DNase1, H3K27ac, H3K36me3
		blood (GM12878)	H3K27ac, H3K36me3, DNase1, H3K4me1, Cmyc
		lung (IMR90)	DNase1, H4K8ac, H2BK12ac, H3K27ac, H3K18ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H4K91ac
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me2, H3K9ac
		skin (NHEK)	H3K36me3, CTCF, DNase1
		NHLF	H3K36me3, H3K27ac, DNase1
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K36me3, H3K27ac, DNase1
		cervix (HeLa-S3)	Cjun, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, TAF1, PolII, H3K79me2, Gabp, Cfos, CTCF, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3, DNase1

		endothelium (HUVEC)	Cmyc, DNase1, PolII, H3K27ac, Max, Cjun, H3K36me3
		A549	DNase1, H3K4me3, H3K36me3
ENSR00001512530 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		breast (HMEC)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001512532 (enhancer)	2	NHLF	H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (K562)	H3K36me3, H3K79me2
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K79me2, H3K36me3
		nervous (NH-A)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001512533 (promoter)	1	embryonic stem cell (H1ESC)	H3K27ac, Yy1, TAF1, H3K4me2, USF1, H3K9ac, H3K4me3, SIX5, Sin3Ak20, p300, H3K36me3, CTCF, PolII, Egr1, Gabp, Jund, TAF7, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K9ac, CTCF, ZBTB7A, Max, H3K27ac, Gabp, USF1, H3K4me3, DNase1, H3K36me3, H3K4me2, PolII, TAF1, HEY1, H3K79me2, Cmyc
		skin (NHDF-AD)	H3K36me3, H3K4me3, DNase1, H3K27ac, H3K4me2, H3K9ac
		muscle (HSMM)	H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	DNase1, H3K4me3, H3K36me3, Cmyc, CTCF, H3K27ac, Gabp, Yy1, PolII, H3K79me2, TAF1, USF1, H3K4me1, H3K4me2, H3K9ac
		lung (IMR90)	DNase1, H3K79me2, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	H3K4me2, H3K9ac, H3K36me3, H3K27ac, H3K4me3, H3K79me2, PolII, BCLAF1, Srf, H2AZ, Cmyc, Yy1, Gabp, ELF1, SIX5, TAF1, DNase1
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac, H3K4me3, H3K27ac
		skin (NHEK)	H3K9ac, H3K27ac, H3K4me1, CTCF, H3K4me2, H3K4me3, DNase1, H3K36me3
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me2, H3K36me3, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me2, H3K4me1, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	H3K36me3, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K36me3, PolII, Gabp, H3K79me2, Ini1, H3K9ac, H3K4me2, H3K27ac, TAF1, Max, H3K4me3, Cmyc
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H4K20me1, H3K9ac, H3K27ac, DNase1
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K27ac, H3K9ac, H3K4me2, H3K4me3, Max
		A549	H3K4me3, H3K36me3, H3K27ac, DNase1, H3K9ac, H3K4me2
ENSR00000275938 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, H3K4me3, H3K4me2, DNase1
		HSMMtube	H3K79me2, DNase1
		blood (K562)	H3K79me2, Max, H3K27ac, DNase1, H3K36me3, H3K4me1
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K79me2, DNase1
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	H3K79me2
		lung (IMR90)	DNase1, H3K79me2, H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1
		NHLF	H3K4me1, DNase1
		Osteobl	H3K4me2, H4K20me1
		blood (DND-41)	H3K36me3, H4K20me1
		cervix (HeLa-S3)	H3K79me2, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

ENSR00000057151 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, H3K36me3, CTCF
		HSMMtube	H3K36me3
		blood (K562)	CTCF
		muscle (HSMM)	H3K36me3
		liver (HepG2)	CTCF, H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3, CTCF
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	CTCF, H3K36me3
		cervix (HeLa-S3)	H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CUL4A	upstream gene variant	73	ENST00000472083	?	?	1
CUL4A	upstream gene variant, downstream gene variant	228	ENST00000488558	?	ENSP00000480367	8
CUL4A	upstream gene variant	901	ENST00000375441	NM_001278513.1	ENSP00000364590	5
CUL4A	upstream gene variant, downstream gene variant	1522	ENST00000470067	?	?	3
CUL4A	upstream gene variant, downstream gene variant	15	ENST00000498562	?	?	8
CUL4A	upstream gene variant, downstream gene variant	410	ENST00000463426	?	?	7
CUL4A	upstream gene variant	1480	ENST00000451881	NM_003589.2	ENSP00000389118	5
CUL4A	upstream gene variant	1544	ENST00000617546	?	ENSP00000481782	5
CUL4A	upstream gene variant, downstream gene variant	233	ENST00000494985	?	?	9
CUL4A	upstream gene variant, downstream gene variant	2324	ENST00000474116	?	?	9
CUL4A	upstream gene variant	2252	ENST00000375440	NM_001008895.2	ENSP00000364589	5
CUL4A	upstream gene variant	1402	ENST00000326335	NM_001278514.1	ENSP00000322132	5
PCID2	downstream gene variant, upstream gene variant	339	ENST00000337344	NM_001127202.2	ENSP00000337405	12
PCID2	downstream gene variant, upstream gene variant	976	ENST00000475433	?	?	10
PCID2	downstream gene variant, upstream gene variant	373	ENST00000375479	NM_018386.3, NM_001127203.2	ENSP00000364628	12
PCID2	downstream gene variant, upstream gene variant	374	ENST00000493650	?	?	13
PCID2	downstream gene variant, upstream gene variant	1590	ENST00000491548	?	?	4
PCID2	downstream gene variant, upstream gene variant	1873	ENST00000484641	?	?	8
PCID2	downstream gene variant, upstream gene variant	301	ENST00000622406	NM_001258212.1	ENSP00000479494	12
PCID2	downstream gene variant, upstream gene variant	579	ENST00000462853	?	?	4
PCID2	downstream gene variant, upstream gene variant	374	ENST00000463102	?	?	13
PCID2	downstream gene variant, upstream gene variant	375	ENST00000462653	?	?	13
PCID2	downstream gene variant, upstream gene variant	1034	ENST00000473462	?	?	12
PCID2	downstream gene variant, upstream gene variant	374	ENST00000375459	?	ENSP00000364608	12
PCID2	downstream gene variant, upstream gene variant	373	ENST00000375477	?	ENSP00000364626	12

PCID2	downstream gene variant, upstream gene variant	374	ENST00000246505	?	ENSP00000246505	12
PCID2	downstream gene variant, upstream gene variant	379	ENST00000375457	NM_001258213.1	ENSP00000364606	12
PCID2	downstream gene variant, upstream gene variant	1035	ENST00000480971	?	?	10
PROZ	downstream gene variant	381	ENST00000342783	NM_001256134.1	ENSP00000344458	10
PROZ	upstream gene variant, downstream gene variant	125	ENST00000493630	?	?	7
PROZ	downstream gene variant	378	ENST00000375547	NM_003891.2	ENSP00000364697	10
RP11-98F14.11	downstream gene variant, upstream gene variant	16	ENST00000600642	?	?	17

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000470067	?	?	1
CUL4A	ENST00000472083	?	?	1
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	1
CUL4A	ENST00000326335	NM_001278514.1	ENSP00000322132	1
CUL4A	ENST00000375440	NM_001008895.2	ENSP00000364589	1
CUL4A	ENST00000617546	?	ENSP00000481782	1
CUL4A	ENST00000451881	NM_003589.2	ENSP00000389118	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CUL4A	ENST00000470067	?	?	6
CUL4A	ENST00000472083	?	?	6
CUL4A	ENST00000375441	NM_001278513.1	ENSP00000364590	30
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PROZ	ENST00000493630	?	?	16
PROZ	ENST00000342783	NM_001256134.1	ENSP00000344458	19

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
PCID2	ENST00000491548	?	1
PCID2	ENST00000493650	?	1