SNiPAcard

Block annotations

Block info

genomic range	chr1:150,273,976-150,514,747
block size	240,772 bp
variant count	69 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.253 [-4.164 – 2.641]	**gene(s) hit or close-by**	AC242988.1, ECM1, FALEC, MRPS21, PRPF3, RPRD2, TARS2
phastCons	μ = 0.096 [0 – 0.998]	**eQTL gene(s)**	C1orf54, CTSS, ECM1, FALEC, HORMAD1, RPRD2, TARS2
GERP++	μ = -0.311 [-5.83 – 4.94]	**potentially regulated gene(s)**	ADAMTSL4, ANP32E, CTSS, MCL1, PRPF3, RP11-458I7.1, RP11-54A4.2, TARS2, VPS45
CADD score	μ = 4.125 [0.007 – 23.2]	**disease gene(s)**	ECM1, ADAMTSL4, PRPF3, TARS2, VPS45

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE	OMIM	MIM:247100
ADAMTSL4	ECTOPIA LENTIS ET PUPILLAE	OMIM	MIM:225200
PRPF3	RETINITIS PIGMENTOSA 18	OMIM	MIM:601414
TARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21	OMIM	MIM:615918
VPS45	NEUTROPENIA, SEVERE CONGENITAL, 5, AUTOSOMAL RECESSIVE	OMIM	MIM:615285
ECM1	Lipoid proteinosis	OrphaNet	OrphaNet:530
ADAMTSL4	isolated ectopia lentis (EL)	OrphaNet	OrphaNet:1885
PRPF3	Retinitis pigmentosa	OrphaNet	OrphaNet:791
VPS45	Recurrent infections-myelofibrosis-nephromegaly syndrome	OrphaNet	OrphaNet:369852

Direct effect on transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
MRPS21	missense variant	ENST00000581066	NM_018997.3	ENSP00000461930	Q/R	cAg/cGg	?	?	1
MRPS21	missense variant	ENST00000614145	NM_031901.5	ENSP00000480129	Q/R	cAg/cGg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
HORMAD1	ENST00000361824	ILMN_1769849	skin	2.23×10^{-5} (p-value)	MuTHER consortium	7
HORMAD1	ENST00000470397					
HORMAD1	ENST00000486497					
HORMAD1	ENST00000322343					
HORMAD1	ENST00000368995					
C1orf54	?	ENSG00000118292	tibial nerve	1.93×10^{-7} (p-value)	GTEx Portal V6	47

FALEC	?	ENSG00000228126	tibial nerve	6.10×10-6 (p-value)	GTEx Portal V6	26
ECM1	?	ENSG00000143369	transformed fibroblasts	3.99×10-10 (p-value)	GTEx Portal V6	66
TARS2	?	ENSG00000143374	transformed fibroblasts	3.66×10-5 (p-value)	GTEx Portal V6	1
ECM1	?	ENSG00000143369	cerebellum	2.37×10-6 (p-value)	GTEx Portal V6	36
ECM1	?	ENSG00000143369	EBV lymphocytes	4.17×10-9 (p-value)	GTEx Portal V6	66
FALEC	?	ENSG00000228126	tibial artery	6.15×10-6 (p-value)	GTEx Portal V6	36
C1orf54	?	ENSG00000118292	tibial artery	8.11×10-6 (p-value)	GTEx Portal V6	3
HORMAD1	?	ENSG00000143452	esophagus mucosa	1.36×10-8 (p-value)	GTEx Portal V6	67
CTSS	?	ENSG00000163131	esophagus mucosa	1.01×10-5 (p-value)	GTEx Portal V6	4
ECM1	?	ENSG00000143369	sun exposed skin	2.08×10-6 (p-value)	GTEx Portal V6	48
FALEC	?	ENSG00000228126	aorta	4.10×10-6 (p-value)	GTEx Portal V6	27
ECM1	?	ENSG00000143369	thyroid	9.15×10-7 (p-value)	GTEx Portal V6	45
HORMAD1	?	ENSG00000143452	subcutaneous adipocytes	2.88×10-5 (p-value)	GTEx Portal V6	1
ECM1	?	ENSG00000143369	blood	1.15×10-6 (p-value)	GTEx Portal V6	23
?	?	ILMN_1702231	blood	3.16×10-5 (p-value)	Westra et al.	1
RPRD2	ENST00000401000	ILMN_3238889	b-cell	7.98×10-4 (p-value)	Fairfax et al.	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036275	1	ENCP00000004323	ADAMTSL4
		ENCP00000004313	PRPF3
		ENCP00000004312	PRPF3
ENCE00000036337	1	ENCP00000004327	MCL1
ENCE00000036132	1	ENCP00000004337	CTSS
		ENCP00000004311	PRPF3
		ENCP00000004291	VPS45
		ENCP00000004300	ANP32E
		ENCP00000004317	TARS2
		ENCP00000004293	VPS45
		ENCP00000004287	RP11-458I7.1
ENCE00000036329	1	ENCP00000004323	ADAMTSL4
ENCE00000036323	1	ENCP00000004325	RP11-54A4.2
		ENCP00000004324	ADAMTSL4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001524644 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (K562)	PolII
		muscle (HSMM)	H3K27ac, DNase1

ENSR00000544743 (promoter)	1	embryonic stem cell (H1ESC)	TAF1, H3K4me2, H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, Jund, TAF7, H3K27ac, Yy1, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, DNase1
		blood (K562)	CTCFL, PolII, H3K4me2, DNase1, H3K4me3, TAF1, H2AZ, Yy1, H3K27ac, Max, PU1, H3K9ac, Cmyc, H3K79me2, HEY1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H2AZ, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		liver (HepG2)	Yy1, PolII, H3K79me2, TAF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me3, H3K4ac, H3K9ac, H4K5ac, H3K4me2, DNase1, H4K8ac, H3K18ac, H3K27ac
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, H3K4me3, H3K79me2, TAF1, PU1, PolII, H2AZ, DNase1, Yy1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Nrf1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Cjun, Max, H3K4me2, H3K4me3, H3K9ac, H3K27ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001586441 (enhancer)	1	cervix (HeLa-S3)	H3K79me2
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
		blood (K562)	H3K27me3, H3K79me2
		muscle (HSMM)	H3K79me2
ENSR00001524646 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00000544755 (promoter flanking region)	2	embryonic stem cell (H1ESC)	DNase1
		liver (HepG2)	H3K4me1, FOXA1
		lung (IMR90)	DNase1, H3K36me3
		blood (K562)	Egr1
		A549	H3K36me3
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, DNase1
		breast (HMEC)	DNase1, H3K27ac, H3K4me2
ENSR00000079774 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII, Gabp, H3K9ac, Yy1, TAF1, H3K4me2, SP1, H3K4me3, DNase1
		HSMMtube	H3K9ac, H3K4me2, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K4me3, DNase1, H3K36me3, H3K27ac, Max, SP2, MEF2A, H3K9ac, Gabp, H3K79me2, HEY1, H2AZ, TAF1, Cfos, PolII, H3K4me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K4me3, H3K4me2, H3K9ac, DNase1
		liver (HepG2)	H3K79me2, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3, H3K36me3, DNase1, Gabp, PolII
		lung (IMR90)	DNase1, H4K8ac, H3K18ac, H3K27ac, H3K4me2, H4K5ac, H3K36me3, H3K4me3, H3K9ac
		blood (GM12878)	ELF1, H3K79me2, H3K4me3, H3K27ac, Cfos, H3K9ac, H3K36me3, Gabp, Yy1, Pbx3, PolII, BCLAF1, MEF2A, H2AZ, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1
		skin (NHEK)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1, H3K27ac
		NHLF	DNase1, H3K4me3, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, Cfos, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00001524653 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K4me2, DNase1
		blood (K562)	H3K27ac, H3K9ac, Jund, PolII, H3K4me2, H3K36me3, DNase1
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, DNase1
		liver (HepG2)	H3K36me3
		blood (GM12878)	PolII, H3K36me3

		lung (IMR90)	DNase1, H3K18ac, H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K4ac, H3K9ac, H4K91ac
		nervous (NH-A)	H3K9ac, H3K4me2, DNase1
		skin (NHEK)	H3K4me1, H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		NHLF	H3K4me3, H3K27ac, H3K36me3
		Osteobl	H3K27ac, H3K4me2, H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K9ac, H3K4me2, H3K27ac, H3K4me1, H3K36me3, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, PolII, DNase1
		A549	H3K36me3
ENSR00000285718 (promoter)	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K27ac, H3K9ac, H2AZ, PolII, H3K4me2, CTCF, H3K4me1, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K36me3, H3K4me1, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, H3K36me3, DNase1
		liver (HepG2)	PolII, Rad21, TAF1, H3K4me1, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
		lung (IMR90)	H3K4me3, H3K9ac, H3K36me3, H3K4me2, DNase1
		blood (GM12878)	Pbx3, PolII, H2AZ, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	H3K9ac, H3K4me2, H3K36me3, DNase1
		NHLF	H3K36me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2, H3K36me3
		blood (DND-41)	H3K4me1, H3K4me2, H3K9ac, H3K4me3, H3K36me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	CTCF, DNase1, Nrf1, H3K9ac, Jund, H3K4me1, PolII
		monocytes (Monocytes-CD14+)	H3K4me3, H3K27me3, DNase1
		endothelium (HUVEC)	H3K4me3, PolII, CTCF, DNase1, Max
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00001524662 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF
		HSMMtube	DNase1, H3K27ac, H3K9ac
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	CTCF, H3K4me1
		lung (IMR90)	DNase1, H3K4me1, H3K18ac, H3K27ac, H4K5ac, H4K91ac
		nervous (NH-A)	DNase1
		skin (NHEK)	CTCF, DNase1

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
PRPF3	3	ENST00000324862 ENST00000496202	hsa-miR-329-3p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC242988.1	downstream gene variant, upstream gene variant	835	ENST00000624245	?	ENSP00000485440	5
ECM1	upstream gene variant, downstream gene variant	976	ENST00000496744	?	?	2
ECM1	upstream gene variant, downstream gene variant	1207	ENST00000490346	?	?	3
ECM1	upstream gene variant, downstream gene variant	2448	ENST00000369049	NM_001202858.1	ENSP00000358045	2
ECM1	upstream gene variant, downstream gene variant	2213	ENST00000369047	NM_004425.3	ENSP00000358043	2
ECM1	upstream gene variant, downstream gene variant	382	ENST00000498579	?	?	3
ECM1	upstream gene variant, downstream gene variant	12	ENST00000346569	NM_022664.2	ENSP00000271630	3
ECM1	upstream gene variant, downstream gene variant	2449	ENST00000470432	?	?	2
FALEC	upstream gene variant, downstream gene variant	4229	ENST00000416894	?	?	2
MRPS21	downstream gene variant	1144	ENST00000614145	NM_031901.5	ENSP00000480129	1

MRPS21	downstream gene variant	1144	ENST00000581066	NM_018997.3	ENSP00000461930	1
PRPF3	upstream gene variant, downstream gene variant	1806	ENST00000324862	NM_004698.2	ENSP00000315379	3
PRPF3	upstream gene variant, downstream gene variant	225	ENST00000470824	?	?	7
PRPF3	upstream gene variant, downstream gene variant	967	ENST00000493553	?	?	7
PRPF3	upstream gene variant, downstream gene variant	1806	ENST00000467329	?	?	7
PRPF3	upstream gene variant, downstream gene variant	3321	ENST00000496202	?	?	3
PRPF3	upstream gene variant, downstream gene variant	3372	ENST00000467514	?	?	5
PRPF3	upstream gene variant	198	ENST00000476970	?	?	1
RPRD2	upstream gene variant, downstream gene variant	1444	ENST00000369067	?	ENSP00000358063	2
RPRD2	upstream gene variant, downstream gene variant	515	ENST00000401000	NM_001297674.1	ENSP00000383785	3
RPRD2	downstream gene variant	3525	ENST00000492220	?	?	2
RPRD2	upstream gene variant, downstream gene variant	1447	ENST00000369068	NM_015203.3	ENSP00000358064	3
TARS2	upstream gene variant	203	ENST00000479372	?	?	2
TARS2	upstream gene variant, downstream gene variant	178	ENST00000369064	NM_025150.4	ENSP00000358060	4
TARS2	upstream gene variant, downstream gene variant	175	ENST00000369054	NM_001271896.1	ENSP00000358050	4
TARS2	upstream gene variant, downstream gene variant	928	ENST00000463555	?	?	3
TARS2	upstream gene variant, downstream gene variant	450	ENST00000460794	?	?	2
TARS2	upstream gene variant, downstream gene variant	928	ENST00000480070	?	?	3
TARS2	upstream gene variant	752	ENST00000466989	?	?	2
TARS2	upstream gene variant, downstream gene variant	460	ENST00000462578	?	?	2
TARS2	upstream gene variant, downstream gene variant	631	ENST00000467982	?	ENSP00000475551	4
TARS2	upstream gene variant, downstream gene variant	181	ENST00000606933	NM_001271895.1	ENSP00000475847	4
TARS2	upstream gene variant, downstream gene variant	196	ENST00000369051	?	ENSP00000358047	4
TARS2	upstream gene variant	192	ENST00000369053	?	?	2
TARS2	downstream gene variant	1160	ENST00000483046	?	?	2
TARS2	upstream gene variant, downstream gene variant	125	ENST00000438568	?	ENSP00000415002	4

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
ECM1	ENST00000369049	NM_001202858.1	ENSP00000358045	T	acA/acG	1
ECM1	ENST00000369047	NM_004425.3	ENSP00000358043	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ECM1	ENST00000346569	NM_022664.2	ENSP00000271630	1
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	4
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	4
PRPF3	ENST00000324862	NM_004698.2	ENSP00000315379	15
PRPF3	ENST00000467329	?	?	10
PRPF3	ENST00000467514	?	?	3

PRPF3	ENST00000476970	?	?	4
PRPF3	ENST00000496202	?	?	8
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	27
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	27
RPRD2	ENST00000369067	?	ENSP00000358063	19
RPRD2	ENST00000492220	?	?	27
TARS2	ENST00000606933	NM_001271895.1	ENSP00000475847	2
TARS2	ENST00000480070	?	?	1
TARS2	ENST00000483046	?	?	1
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	2
TARS2	ENST00000369051	?	ENSP00000358047	2
TARS2	ENST00000467982	?	ENSP00000475551	2
TARS2	ENST00000438568	?	ENSP00000415002	2
TARS2	ENST00000463555	?	?	1
TARS2	ENST00000369054	NM_001271896.1	ENSP00000358050	2

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ECM1	ENST00000369049	NM_001202858.1	ENSP00000358045	1
ECM1	ENST00000369047	NM_004425.3	ENSP00000358043	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ECM1	ENST00000496744	?	1
ECM1	ENST00000470432	?	2
ECM1	ENST00000498579	?	1
FALEC	ENST00000416894	?	1
RPRD2	ENST00000492220	?	1