SNiPAcard

Block annotations

Block info

genomic range	chr1:150,267,798-150,511,628
block size	243,831 bp
variant count	138 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.549 [-3.729 – 1.45]	**gene(s) hit or close-by**	AC242988.1, ECM1, FALEC, MRPS21, PRPF3, RPRD2, TARS2
phastCons	μ = 0.061 [0 – 1]	**eQTL gene(s)**	ADAMTSL4, CDC42SE1, ECM1, ENSA, MRPS21, RP11-54A4.2, RPRD2, TARS2
GERP++	μ = -0.256 [-7.72 – 4.17]	**potentially regulated gene(s)**	ANXA9, APH1A, MCL1, RP11-235D19.2, VPS45
CADD score	μ = 3.547 [0.014 – 16.23]	**disease gene(s)**	ADAMTSL4, TARS2, ECM1, PRPF3, VPS45

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ADAMTSL4	ECTOPIA LENTIS ET PUPILLAE	OMIM	MIM:225200
TARS2	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 21	OMIM	MIM:615918
ECM1	LIPOID PROTEINOSIS OF URBACH AND WIETHE	OMIM	MIM:247100
PRPF3	RETINITIS PIGMENTOSA 18	OMIM	MIM:601414
VPS45	NEUTROPENIA, SEVERE CONGENITAL, 5, AUTOSOMAL RECESSIVE	OMIM	MIM:615285
ADAMTSL4	isolated ectopia lentis (EL)	OrphaNet	OrphaNet:1885
ECM1	Lipoid proteinosis	OrphaNet	OrphaNet:530
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)
ECM1	missense variant	ENST00000346569	NM_022664.2	ENSP00000271630	2	2			2
ECM1	missense variant	ENST00000369049	NM_001202858.1	ENSP00000358045	2	2			2
ECM1	missense variant	ENST00000369047	NM_004425.3	ENSP00000358043	2	2			2

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
MRPS21	ENST00000581066	ILMN_1660292	blood	1.51×10^{-8} (p-value)	MuTHER consortium	32
			adipocyte	2.23×10^{-20} (p-value)	MuTHER consortium	32
MRPS21	ENST00000581066	ILMN_1655765	blood	8.19×10^{-7} (p-value)	MuTHER consortium	32

MRPS21	ENST00000614145					
CDC42SE1	ENST00000540998	ILMN_2349138	blood	5.88×10-5 (p-value)	MuTHER consortium	2
CDC42SE1	ENST00000483763					
CDC42SE1	ENST00000357235					
CDC42SE1	ENST00000439374					
CDC42SE1	ENST00000483763	ILMN_1769027	blood	5.62×10-6 (p-value)	MuTHER consortium	32
CDC42SE1	ENST00000357235					
CDC42SE1	ENST00000540998					
CDC42SE1	ENST00000439374					
TARS2	?	ENSG00000143374	visceral adipocytes	1.00×10-6 (p-value)	GTEx Portal V6	125
RPRD2	?	ENSG00000163125	blood	9.70×10-6 (p-value)	GTEx Portal V6	56
TARS2	?	ENSG00000143374	breast	5.13×10-7 (p-value)	GTEx Portal V6	126
RP11-54A4.2	?	ENSG00000237781	tibial artery	4.92×10-8 (p-value)	GTEx Portal V6	131
ADAMTSL4	?	ENSG00000143382	tibial artery	4.27×10-7 (p-value)	GTEx Portal V6	80
TARS2	?	ENSG00000143374	tibial artery	1.42×10-6 (p-value)	GTEx Portal V6	97
RP11-54A4.2	?	ENSG00000237781	esophagus mucosa	1.06×10-9 (p-value)	GTEx Portal V6	134
ADAMTSL4	?	ENSG00000143382	esophagus mucosa	3.79×10-11 (p-value)	GTEx Portal V6	134
ECM1	?	ENSG00000143369	esophagus mucosa	8.32×10-6 (p-value)	GTEx Portal V6	10
?	?	ILMN_1702231	blood	1.24×10-6 (p-value)	Westra et al.	33
RPRD2	?	ENSG00000163125	testis	2.07×10-8 (p-value)	GTEx Portal V6	134
TARS2	?	ENSG00000143374	skeletal muscle	2.84×10-8 (p-value)	GTEx Portal V6	132
TARS2	?	ENSG00000143374	unexposed skin	4.82×10-8 (p-value)	GTEx Portal V6	129
ADAMTSL4	?	ENSG00000143382	aorta	1.07×10-7 (p-value)	GTEx Portal V6	132
RP11-54A4.2	?	ENSG00000237781	aorta	1.43×10-5 (p-value)	GTEx Portal V6	3
TARS2	?	ENSG00000143374	subcutaneous adipocytes	2.47×10-7 (p-value)	GTEx Portal V6	129
RPRD2	ENST00000401000	ILMN_3238889	monocyte	9.96×10-6 (p-value)	Fairfax et al.	3
			b-cell	6.13×10-6 (p-value)	Fairfax et al.	3
MRPS21	ENST00000581066	ILMN_1660292	monocyte	2.64×10-14 (p-value)	Fairfax et al.	2
			b-cell	1.15×10-4 (p-value)	Fairfax et al.	2
RPRD2	?	ENSG00000163125	lung	1.48×10-5 (p-value)	GTEx Portal V6	42
ENSA	?	ENSG00000143420	liver	5.48×10-6 (p-value)	GTEx Portal V6	1
ADAMTSL4	?	ENSG00000143382	transformed fibroblasts	8.76×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000036149	1	ENCP00000004292	VPS45
		ENCP00000004293	VPS45
ENCE00000036228	2	ENCP00000004307	APH1A

ENCE00000036339	1	ENCP00000004360	ANXA9
		ENCP00000004327	MCL1
ENCE00000036433	1	ENCP00000004346	RP11-235D19.2

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001524644 (open chromatin region)	2	embryonic stem cell (H1ESC)	H3K36me3, PolII
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		blood (K562)	PolII
		muscle (HSMM)	H3K27ac, DNase1
ENSR00001586440 (CTCF binding site)	1	embryonic stem cell (H1ESC)	H3K4me2, H3K4me3
		HSMMtube	H3K79me2, H3K4me2
		blood (K562)	H3K27ac, H3K9ac, H3K79me2, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, H3K9ac, H3K4me2
		muscle (HSMM)	H3K79me2, H3K4me3, H3K4me2
		liver (HepG2)	H3K79me2, H3K4me2, H3K9ac, H3K4me3
		blood (GM12878)	H3K79me2, H3K4me3, H3K27ac, H3K9ac, H3K36me3
		lung (IMR90)	H3K79me2, H3K4me2, H3K36me3, H3K4me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, H3K4me3
		NHLF	H3K4me3, H3K27ac
		Osteobl	H3K4me3, H3K4me2
		blood (DND-41)	H3K4me3, H3K36me3, H3K4me1, H3K4me2, H3K27ac, H3K9ac
		breast (HMEC)	H3K4me3, H3K4me2
		cervix (HeLa-S3)	H3K79me2, H3K4me3, H3K4me2, PolII, H3K9ac, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
ENSR00000544743 (promoter)	2	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
ENSR00001586444 (enhancer)	1	cervix (HeLa-S3)	H3K79me2
		monocytes (Monocytes-CD14+)	H4K20me1
		liver (HepG2)	H3K79me2
		blood (GM12878)	H3K79me2
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K79me2
ENSR00001524646 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1, FOXA1
		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00001586446 (enhancer)	1	liver (HepG2)	H3K79me2, H3K4me1
		blood (DND-41)	H3K36me3
ENSR00001586448	1	liver (HepG2)	H3K79me2, H3K4me1, H3K4me2, H3K27ac

(enhancer)		blood (GM12878)	H3K79me2
		blood (DND-41)	H3K36me3
ENSR00000544752	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
(open chromatin region)		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00000544755	2	embryonic stem cell (H1ESC)	DNase1
(promoter flanking region)		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
ENSR00000285713	1	embryonic stem cell (H1ESC)	CTCF, Rad21
(promoter flanking region)		Osteobl	CTCF
		blood (K562)	DNase1, Rad21, H2AZ, CTCF
		skin (NHDF-AD)	CTCF, DNase1
		breast (HMEC)	CTCF
		muscle (HSMM)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		endothelium (HUVEC)	H3K36me3, CTCF
		liver (HepG2)	CTCF, H3K4me2, Rad21, H2AZ
		lung (IMR90)	CTCF
		skin (NHEK)	CTCF, DNase1
ENSR00000285718	1	embryonic stem cell (H1ESC)	H3K36me3, H3K27me3, CTCF, Rad21, TAF1, H3K4me2, H3K4me3, DNase1
(promoter)		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	3	NHLF	DNase1
(promoter flanking region)		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
ENSR00001586454	1	NHLF	DNase1
(CTCF binding site)		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		HSMMtube	H3K9ac, H3K27ac, DNase1
		blood (K562)	CTCF
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	H3K27ac, DNase1
		cervix (HeLa-S3)	CTCF, DNase1
		liver (HepG2)	CTCF
		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	2	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	893	ENST00000624245	?	ENSP00000485440	4
ECM1	upstream gene variant, downstream gene variant	657	ENST00000498579	?	?	6
ECM1	upstream gene variant, downstream gene variant	727	ENST00000490346	?	?	4
ECM1	upstream gene variant, downstream gene variant	682	ENST00000346569	NM_022664.2	ENSP00000271630	6
ECM1	upstream gene variant, downstream gene variant	651	ENST00000369049	NM_001202858.1	ENSP00000358045	6
ECM1	upstream gene variant, downstream gene variant	637	ENST00000470432	?	?	6
ECM1	upstream gene variant, downstream gene variant	660	ENST00000369047	NM_004425.3	ENSP00000358043	6
ECM1	upstream gene variant, downstream gene variant	327	ENST00000496744	?	?	7
FALEC	upstream gene variant, downstream gene variant	57	ENST00000416894	?	?	7
MRPS21	downstream gene variant	86	ENST00000581066	NM_018997.3	ENSP00000461930	7
MRPS21	downstream gene variant	86	ENST00000614145	NM_031901.5	ENSP00000480129	7
PRPF3	downstream gene variant	266	ENST00000476970	?	?	4
PRPF3	upstream gene variant, downstream gene variant	1435	ENST00000467329	?	?	5
PRPF3	upstream gene variant, downstream gene variant	771	ENST00000324862	NM_004698.2	ENSP00000315379	9
PRPF3	upstream gene variant, downstream gene variant	774	ENST00000496202	?	?	8
PRPF3	downstream gene variant	2184	ENST00000467514	?	?	3
PRPF3	upstream gene variant, downstream gene variant	3359	ENST00000493553	?	?	6
PRPF3	upstream gene variant, downstream gene variant	313	ENST00000470824	?	?	7
RPRD2	upstream gene variant, downstream gene variant	953	ENST00000401000	NM_001297674.1	ENSP00000383785	7
RPRD2	upstream gene variant, downstream gene variant	1366	ENST00000492220	?	?	3
RPRD2	upstream gene variant	1011	ENST00000369067	?	ENSP00000358063	3
RPRD2	upstream gene variant, downstream gene variant	1014	ENST00000369068	NM_015203.3	ENSP00000358064	4
TARS2	upstream gene variant, downstream gene variant	1964	ENST00000438568	?	ENSP00000415002	3
TARS2	upstream gene variant, downstream gene variant	2031	ENST00000369053	?	?	2
TARS2	upstream gene variant, downstream gene variant	293	ENST00000483046	?	?	6
TARS2	upstream gene variant, downstream gene variant	167	ENST00000369054	NM_001271896.1	ENSP00000358050	3
TARS2	upstream gene variant, downstream gene variant	149	ENST00000480070	?	?	3
TARS2	upstream gene variant, downstream gene variant	149	ENST00000463555	?	?	3
TARS2	upstream gene variant, downstream gene variant	2042	ENST00000479372	?	?	2
TARS2	upstream gene variant, downstream gene variant	259	ENST00000462578	?	?	6
TARS2	upstream gene variant, downstream gene variant	1759	ENST00000466989	?	?	7
TARS2	upstream gene variant, downstream gene variant	249	ENST00000460794	?	?	6
TARS2	upstream gene variant, downstream gene variant	242	ENST00000467982	?	ENSP00000475551	3
TARS2	upstream gene variant, downstream gene variant	141	ENST00000606933	NM_001271895.1	ENSP00000475847	3
TARS2	upstream gene variant, downstream gene variant	2035	ENST00000369051	?	ENSP00000358047	3

TARS2	upstream gene variant, downstream gene variant	2017	ENST00000369064	NM_025150.4	ENSP00000358060	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	7
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	7
PRPF3	ENST00000470824	?	?	5
PRPF3	ENST00000467514	?	?	2
PRPF3	ENST00000476970	?	?	4
PRPF3	ENST00000496202	?	?	4
PRPF3	ENST00000467329	?	?	13
PRPF3	ENST00000324862	NM_004698.2	ENSP00000315379	15
RPRD2	ENST00000369067	?	ENSP00000358063	47
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	61
RPRD2	ENST00000492220	?	?	62
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	61
TARS2	ENST00000467982	?	ENSP00000475551	15
TARS2	ENST00000369053	?	?	1
TARS2	ENST00000480070	?	?	13
TARS2	ENST00000460794	?	?	6
TARS2	ENST00000606933	NM_001271895.1	ENSP00000475847	15
TARS2	ENST00000369051	?	ENSP00000358047	15
TARS2	ENST00000463555	?	?	13
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	15
TARS2	ENST00000462578	?	?	6
TARS2	ENST00000466989	?	?	1
TARS2	ENST00000483046	?	?	1
TARS2	ENST00000438568	?	ENSP00000415002	15
TARS2	ENST00000369054	NM_001271896.1	ENSP00000358050	15

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MRPS21	ENST00000614145	NM_031901.5	ENSP00000480129	2
MRPS21	ENST00000581066	NM_018997.3	ENSP00000461930	2
RPRD2	ENST00000401000	NM_001297674.1	ENSP00000383785	2
RPRD2	ENST00000369068	NM_015203.3	ENSP00000358064	1
TARS2	ENST00000369051	?	ENSP00000358047	1
TARS2	ENST00000369064	NM_025150.4	ENSP00000358060	1
TARS2	ENST00000438568	?	ENSP00000415002	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ECM1	ENST00000498579	?	1
ECM1	ENST00000470432	?	2
PRPF3	ENST00000470824	?	1
RPRD2	ENST00000492220	?	1
TARS2	ENST00000479372	?	1