SNiPAcard

Block annotations

Block info

genomic range	chr1:45,954,632-46,252,418
block size	297,787 bp
variant count	44 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.187 [-2.036 – 1.388]	**gene(s) hit or close-by**	AL355480.1, AL355480.3, AL355480.4, CCDC163P, CCDC17, GPBP1L1, IPP, MAST2, MMACHC, NASP, PRDX1, RP11-630I5.1, RP11-767N6.2, RP11-767N6.7, RPS15AP10, TESK2, TMEM69
phastCons	μ = 0.085 [0 – 0.978]	**eQTL gene(s)**	MAST2, MMACHC, PIK3R3, PRDX1, TMEM69
GERP++	μ = -0.131 [-3.98 – 2.7]	**potentially regulated gene(s)**	AKR1A1, GPBP1L1, HPDL, MAST2, PIK3R3, TESK2, TOE1
CADD score	μ = 3.765 [0.207 – 17.61]	**disease gene(s)**	MMACHC

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Blood metabolite levels	$<5.00\times10^{-11}$	GWAS Catalog	24816252	1
erythritol	4.99×10^{-11}	Metabolomics GWAS Server	24816252	7

Disease gene annotation

gene	trait	source DB	source entry/link
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	OMIM	MIM:277400
MMACHC	HOMOCYSTEINEMIA	OMIM	MIM:603174
MMACHC	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE	DECIPHER	MIM:277400
MMACHC	Methylmalonic acidemia with homocystinuria, type cblC	OrphaNet	OrphaNet:79282

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TMEM69	?	ENSG00000159596	transformed fibroblasts	1.37×10^{-8} (p-value)	GTEx Portal V6	41
MAST2	?	ENSG00000086015	transformed fibroblasts	3.76×10^{-8} (p-value)	GTEx Portal V6	40
PIK3R3	?	ENSG00000117461	transformed fibroblasts	2.79×10^{-5} (p-value)	GTEx Portal V6	1
MAST2	?	ENSG00000086015	tibial artery	7.72×10^{-10} (p-value)	GTEx Portal V6	41
PRDX1	?	ENSG00000117450	tibial artery	8.10×10^{-7} (p-value)	GTEx Portal V6	27
PRDX1	?	ENSG00000117450	thyroid	1.86×10^{-10} (p-value)	GTEx Portal V6	40
MMACHC	?	ENSG00000132763	subcutaneous adipocytes	4.36×10^{-8} (p-value)	GTEx Portal V6	38
MAST2	?	ENSG00000086015	left ventricle	4.91×10^{-7} (p-value)	GTEx Portal V6	36
MMACHC	ENST00000616135	ILMN_1803005	blood	1.08×10^{-6} (p-value)	MuTHER consortium	8
MMACHC	ENST00000401061		adipocyte	4.91×10^{-10} (p-value)	MuTHER consortium	8

MMACHC	ENST00000477188					
PRDX1	ENST00000372079	ILMN_2366388	monocyte	6.74×10-4 (p-value)	Fairfax et al.	1
PRDX1	ENST00000319248					
PRDX1	ENST00000262746					
MAST2	?	ENSG00000086015	tibial nerve	4.13×10-6 (p-value)	GTEx Portal V6	4

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000017982	1	ENCP00000002314	GPBP1L1
		ENCP00000002293	TESK2
		ENCP00000002331	MAST2
		ENCP00000002305	AKR1A1
		ENCP00000002336	MAST2
ENCE00000018297	1	ENCP00000002340	PIK3R3
ENCE00000017914	1	ENCP00000002318	GPBP1L1
		ENCP00000002286	HPDL
ENCE00000017958	1	ENCP00000002291	TOE1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000669883 (promoter)	1	embryonic stem cell (H1ESC)	H3K79me2, H4K5ac, H3K36me3, H3K27me3, CTCF, PolII, Rad21, TAF7, Sin3Ak20, H3K27ac, Yy1, TAF1, H3K4me2, SP1, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K4me2, H3K4me3, H2AZ, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, ELF1, PolII, GTF2B, H4K20me1, H3K4me2, H3K36me3, DNase1, H3K4me3, Cmyc, Gabp, Yy1, Egr1, H3K27ac, Max, E2F6, H3K9ac
		skin (NHDF-AD)	H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H2AZ, H3K79me2, H3K4me3, H3K27ac
		liver (HepG2)	H3K4me3, DNase1, H3K27ac, H3K9ac, Gabp, Yy1, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H2AZ, H3K4me2
		blood (GM12878)	H3K9ac, H3K4me2, Cmyc, Max, PolII, H2AZ, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac
		lung (IMR90)	DNase1, H3K79me2, H3K18ac, H3K4me2, H3K4me3, H3K9ac
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K36me3, DNase1
		skin (NHEK)	H3K4me1, H3K27ac, H3K9ac, H3K4me2, H3K4me3, DNase1
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K9ac, H3K4me3, H3K36me3, H4K20me1, H3K27ac, H3K4me1, H3K4me2
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, H3K79me2, Gabp, PolII, H3K36me3
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	DNase1, Max, H3K4me3, H3K4me2, H3K9ac, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00001519843 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (DND-41)	H3K36me3
		blood (K562)	H3K79me2, H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000535923 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, Yy1
		HSMMtube	H3K79me2

		Osteobl	H3K4me2
		blood (K562)	H3K79me2, H4K20me1
		blood (DND-41)	H4K20me1, H3K36me3
		muscle (HSMM)	H3K79me2
		cervix (HeLa-S3)	H3K79me2, H3K36me3
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K4me2
		liver (HepG2)	H4K20me1, H3K79me2, H3K4me1
		lung (IMR90)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
ENSR00000282137 (enhancer)	1	blood (K562)	DNase1
ENSR00000165101 (enhancer)	1	HSMMtube	H3K27me3
		blood (DND-41)	H3K4me1
		blood (K562)	H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AL355480.1	downstream gene variant, upstream gene variant	2577	ENST00000629041	?	?	2
AL355480.3	downstream gene variant, upstream gene variant	2250	ENST00000630128	?	?	2
AL355480.4	downstream gene variant, upstream gene variant	277	ENST00000626823	?	?	3
CCDC163P	downstream gene variant	4906	ENST00000626177	?	ENSP00000485784	1
CCDC163P	downstream gene variant	4966	ENST00000625766	?	ENSP00000486505	1
CCDC163P	downstream gene variant	4977	ENST00000629009	?	?	1
CCDC17	downstream gene variant, upstream gene variant	1512	ENST00000445048	?	ENSP00000411335	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000372044	?	ENSP00000361114	3
CCDC17	downstream gene variant, upstream gene variant	1642	ENST00000479529	?	ENSP00000437072	3
CCDC17	downstream gene variant, upstream gene variant	1334	ENST00000421127	NM_001190182.1	ENSP00000389415	3
CCDC17	downstream gene variant, upstream gene variant	337	ENST00000464739	?	?	4
CCDC17	downstream gene variant, upstream gene variant	1400	ENST00000528266	NM_001114938.2	ENSP00000432172	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000482416	?	?	3
CCDC17	downstream gene variant, upstream gene variant	1333	ENST00000491755	?	?	3
CCDC17	downstream gene variant, upstream gene variant	1624	ENST00000525599	?	?	4
GPBP1L1	downstream gene variant	4062	ENST00000480941	?	?	1
GPBP1L1	downstream gene variant	611	ENST00000290795	?	ENSP00000290795	1
GPBP1L1	downstream gene variant	611	ENST00000479235	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	309	ENST00000496278	?	?	3
GPBP1L1	downstream gene variant	3949	ENST00000488278	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	602	ENST00000467032	?	?	2
GPBP1L1	downstream gene variant	4330	ENST00000460859	?	?	1
GPBP1L1	upstream gene variant	1222	ENST00000498128	?	?	1
GPBP1L1	downstream gene variant	2895	ENST00000468724	?	?	1
GPBP1L1	downstream gene variant, upstream gene variant	2287	ENST00000495616	?	?	3
GPBP1L1	downstream gene variant	613	ENST00000355105	NM_021639.4	ENSP00000347224	1

GPBP1L1	downstream gene variant	1535	ENST00000487436	?	?	1
GPBP1L1	downstream gene variant	4209	ENST00000493083	?	?	1
GPBP1L1	downstream gene variant	4081	ENST00000480083	?	?	1
IPP	downstream gene variant, upstream gene variant	863	ENST00000495072	?	?	3
IPP	upstream gene variant	1171	ENST00000359942	NM_001145349.1	ENSP00000353024	1
IPP	downstream gene variant	863	ENST00000461718	?	?	2
IPP	downstream gene variant, upstream gene variant	906	ENST00000396478	NM_005897.2	ENSP00000379739	4
MAST2	upstream gene variant	241	ENST00000470809	?	?	3
MMACHC	downstream gene variant	1508	ENST00000477188	?	?	1
MMACHC	downstream gene variant	948	ENST00000616135	?	ENSP00000478859	1
NASP	upstream gene variant, downstream gene variant	2319	ENST00000527932	?	?	4
NASP	downstream gene variant	2628	ENST00000527359	?	ENSP00000436790	2
NASP	downstream gene variant	2571	ENST00000528238	?	ENSP00000432289	2
NASP	downstream gene variant	3917	ENST00000525515	?	ENSP00000436939	2
NASP	upstream gene variant, downstream gene variant	467	ENST00000530073	?	?	3
NASP	downstream gene variant	2903	ENST00000437901	?	ENSP00000400792	2
NASP	downstream gene variant	3447	ENST00000464190	?	?	1
NASP	downstream gene variant	2903	ENST00000437362	?	ENSP00000388010	2
NASP	downstream gene variant	2594	ENST00000527470	?	ENSP00000437241	1
NASP	downstream gene variant	2463	ENST00000534101	?	?	1
NASP	upstream gene variant, downstream gene variant	427	ENST00000481782	?	?	4
NASP	downstream gene variant	464	ENST00000351223	NM_152298.3	ENSP00000255121	2
NASP	upstream gene variant, downstream gene variant	498	ENST00000531612	?	ENSP00000437116	3
NASP	downstream gene variant	465	ENST00000372052	?	ENSP00000361122	2
NASP	downstream gene variant	3595	ENST00000528084	?	ENSP00000435363	1
NASP	downstream gene variant	465	ENST00000472408	?	?	2
NASP	upstream gene variant, downstream gene variant	464	ENST00000534450	?	ENSP00000434240	3
NASP	downstream gene variant	1763	ENST00000350030	NM_002482.3	ENSP00000255120	1
NASP	downstream gene variant	2632	ENST00000529333	?	ENSP00000436388	1
NASP	downstream gene variant	1753	ENST00000537798	NM_001195193.1	ENSP00000438871	1
NASP	downstream gene variant	991	ENST00000531532	?	ENSP00000436778	1
NASP	downstream gene variant	2155	ENST00000470768	?	ENSP00000436924	1
NASP	downstream gene variant	3729	ENST00000530840	?	ENSP00000435548	1
NASP	downstream gene variant	2680	ENST00000453748	?	ENSP00000403157	2
PRDX1	downstream gene variant, upstream gene variant	1419	ENST00000319248	NM_181697.2	ENSP00000361152	2
PRDX1	downstream gene variant, upstream gene variant	3540	ENST00000483583	?	?	2
PRDX1	downstream gene variant, upstream gene variant	1408	ENST00000262746	NM_181696.2, NM_002574.3, NM_001202431.1	ENSP00000262746	2

PRDX1	downstream gene variant, upstream gene variant	2360	ENST00000424390 ?	ENSP00000389047	3
PRDX1	downstream gene variant, upstream gene variant	1423	ENST00000372079 ?	ENSP00000361150	2
PRDX1	downstream gene variant, upstream gene variant	2494	ENST00000447184 ?	ENSP00000407034	3
RP11-630I5.1	upstream gene variant, downstream gene variant	1526	ENST00000463059 ?	?	4
RP11-767N6.2	upstream gene variant	895	ENST00000437848 ?	?	3
RP11-767N6.7	upstream gene variant	730	ENST00000430643 ?	?	1
RPS15AP10	downstream gene variant	394	ENST00000432472 ?	?	1
TMEM69	downstream gene variant	1819	ENST00000496366 ?	?	3
TMEM69	downstream gene variant	1221	ENST00000372025 NM_016486.3	ENSP00000361095	3

Putative effect on transcript

Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
CCDC17	ENST00000479529	?	ENSP00000437072	T	acA/acG	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CCDC17	ENST00000528266	NM_001114938.2	ENSP00000432172	1
CCDC17	ENST00000491755	?	?	1
CCDC17	ENST00000421127	NM_001190182.1	ENSP00000389415	1
CCDC17	ENST00000445048	?	ENSP00000411335	1
CCDC17	ENST00000372044	?	ENSP00000361114	1
CCDC17	ENST00000482416	?	?	1
GPBP1L1	ENST00000480083	?	?	5
GPBP1L1	ENST00000488278	?	?	5
GPBP1L1	ENST00000493083	?	?	5
GPBP1L1	ENST00000498128	?	?	3
GPBP1L1	ENST00000460859	?	?	5
GPBP1L1	ENST00000468724	?	?	1
GPBP1L1	ENST00000290795	?	ENSP00000290795	10
GPBP1L1	ENST00000479235	?	?	1
GPBP1L1	ENST00000487436	?	?	1
GPBP1L1	ENST00000480941	?	?	5
GPBP1L1	ENST00000355105	NM_021639.4	ENSP00000347224	10
IPP	ENST00000396478	NM_005897.2	ENSP00000379739	10
IPP	ENST00000359942	NM_001145349.1	ENSP00000353024	13
IPP	ENST00000461718	?	?	1
IPP	ENST00000495072	?	?	2
MMACHC	ENST00000616135	?	ENSP00000478859	1
MMACHC	ENST00000477188	?	?	1

NASP	ENST00000481782	?	?	1
NASP	ENST00000528084	?	ENSP00000435363	3
NASP	ENST00000351223	NM_152298.3	ENSP00000255121	5
NASP	ENST00000530840	?	ENSP00000435548	2
NASP	ENST00000453748	?	ENSP00000403157	4
NASP	ENST00000531612	?	ENSP00000437116	1
NASP	ENST00000437901	?	ENSP00000400792	4
NASP	ENST00000525515	?	ENSP00000436939	4
NASP	ENST00000470768	?	ENSP00000436924	3
NASP	ENST00000372052	?	ENSP00000361122	5
NASP	ENST00000472408	?	?	1
NASP	ENST00000528238	?	ENSP00000432289	4
NASP	ENST00000534450	?	ENSP00000434240	1
NASP	ENST00000527359	?	ENSP00000436790	4
NASP	ENST00000531532	?	ENSP00000436778	2
NASP	ENST00000537798	NM_001195193.1	ENSP00000438871	5
NASP	ENST00000350030	NM_002482.3	ENSP00000255120	5
NASP	ENST00000437362	?	ENSP00000388010	4
NASP	ENST00000629893	?	ENSP00000486336	1
NASP	ENST00000534101	?	?	3
NASP	ENST00000529333	?	ENSP00000436388	3
NASP	ENST00000527470	?	ENSP00000437241	3
NASP	ENST00000464190	?	?	3
NASP	ENST00000530073	?	?	2
PRDX1	ENST00000319248	NM_181697.2	ENSP00000361152	1
PRDX1	ENST00000262746	NM_181696.2, NM_002574.3, NM_001202431.1	ENSP00000262746	1
PRDX1	ENST00000372079	?	ENSP00000361150	1
RP11-767N6.7	ENST00000430643	?	?	2
TESK2	ENST00000372086	NM_007170.2	ENSP00000361158	1
TESK2	ENST00000486676	?	?	1

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MMACHC	ENST00000401061	NM_015506.2	ENSP00000383840	2
NASP	ENST00000537798	NM_001195193.1	ENSP00000438871	1
NASP	ENST00000350030	NM_002482.3	ENSP00000255120	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NASP	ENST00000530073	?	1