SNiPAcard

Block annotations

Block info

genomic range	chr2:71,292,522-71,327,352
block size	34,831 bp
variant count	8 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.943 [-2.25 – 0.896]	**gene(s) hit or close-by**	AC007040.8, NAGK, RP11-467P9.1
phastCons	μ = 0.011 [0 – 0.061]	**eQTL gene(s)**	MCEE, NAGK, OR7E62P, OR7E91P
GERP++	μ = -0.097 [-2.23 – 1.49]	**potentially regulated gene(s)**	AC007040.6, AC007040.8, CD207, MPHOSPH10, NAGK, ZNF638
CADD score	μ = 3.759 [0.492 – 7.392]	**disease gene(s)**	MCEE, CD207

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY	DECIPHER	MIM:251120
MCEE	METHYLMALONYL-CoA EPIMERASE DEFICIENCY	OMIM	MIM:251120
CD207	BIRBECK GRANULE DEFICIENCY	OMIM	MIM:613393
MCEE	Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency	OrphaNet	OrphaNet:308425

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
NAGK	?	ENSG00000124357	muscularis mucosae	2.31×10^{-7} (p-value)	GTEx Portal V6	3
NAGK	?	ENSG00000124357	lung	8.00×10^{-13} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	spleen	1.41×10^{-7} (p-value)	GTEx Portal V6	2
NAGK	?	ENSG00000124357	tibial nerve	1.12×10^{-8} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	blood	8.69×10^{-24} (p-value)	GTEx Portal V6	8
MCEE	?	ENSG00000124370	esophagus mucosa	4.93×10^{-8} (p-value)	GTEx Portal V6	8
OR7E91P	?	ENSG00000205847	esophagus mucosa	2.33×10^{-8} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	esophagus mucosa	8.14×10^{-29} (p-value)	GTEx Portal V6	8
NAGK	?	ENSG00000124357	sun exposed skin	1.07×10^{-9} (p-value)	GTEx Portal V6	8
OR7E62P	?	ENSG00000234896	sun exposed skin	2.09×10^{-6} (p-value)	GTEx Portal V6	1
NAGK	?	ENSG00000124357	subcutaneous adipocytes	6.67×10^{-7} (p-value)	GTEx Portal V6	1
MCEE	ENST00000413592	ILMN_1735347	blood	2.66×10^{-6} (p-value)	Westra et al.	3
MCEE	ENST00000244217					
MCEE	ENST00000462609					
NAGK	?	ENSG00000124357	EBV lymphocytes	2.53×10^{-6} (p-value)	GTEx Portal V6	1
NAGK	?	ENSG00000124357	blood	2.95×10^{-3} (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000251577	1	ENCP00000026542	MPHOSPH10
		ENCP00000026554	ZNF638
		ENCP00000026534	NAGK
		ENCP00000026508	CD207
		ENCP00000026522	AC007040.6
		ENCP00000026535	NAGK
		ENCP00000026528	AC007040.8
		ENCP00000026553	ZNF638
		ENCP00000026540	MPHOSPH10
		ENCP00000026541	MPHOSPH10

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000592557 (CTCF binding site)	1	embryonic stem cell (H1ESC)	DNase1, Rad21, CTCF, Yy1
		HSMMtube	CTCF
		blood (K562)	Rad21, H2AZ, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, H3K4me1, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Yy1, Rad21, CTCF, DNase1
		nervous (NH-A)	CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	CTCF, H3K4me2
		blood (DND-41)	CTCF, H3K27me3
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	H3K4me3, CTCF

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC007040.8	upstream gene variant	649	ENST00000434990	?	?	1
NAGK	upstream gene variant	2916	ENST00000531934	?	ENSP00000436326	1
NAGK	upstream gene variant	2909	ENST00000455662	?	ENSP00000389087	1
NAGK	upstream gene variant	3239	ENST00000493102	?	?	1
NAGK	upstream gene variant	3586	ENST00000489309	?	?	1
NAGK	upstream gene variant	3258	ENST00000480411	?	?	1
NAGK	upstream gene variant	2886	ENST00000613852	NM_017567.4	ENSP00000477639	1
NAGK	upstream gene variant	3255	ENST00000450272	?	ENSP00000400941	1
NAGK	upstream gene variant	3258	ENST00000465105	?	?	1
NAGK	upstream gene variant	3230	ENST00000484984	?	?	1
NAGK	upstream gene variant	3196	ENST00000418807	?	ENSP00000396070	1
NAGK	upstream gene variant	2924	ENST00000455197	?	ENSP00000408550	1

NAGK	upstream gene variant	3258	ENST00000475709	?	?	1
NAGK	upstream gene variant, downstream gene variant	233	ENST00000497690	?	?	2
NAGK	upstream gene variant	4908	ENST00000464638	?	?	1
NAGK	upstream gene variant	3230	ENST00000244204	?	ENSP00000244204	1
NAGK	upstream gene variant	3273	ENST00000472519	?	?	1
NAGK	upstream gene variant	3227	ENST00000443938	?	ENSP00000411846	1
NAGK	upstream gene variant	1433	ENST00000478659	?	?	1
NAGK	upstream gene variant	3270	ENST00000533981	?	ENSP00000435207	1
NAGK	upstream gene variant	3200	ENST00000443872	?	ENSP00000404531	1
NAGK	upstream gene variant	939	ENST00000529236	?	ENSP00000431204	1
NAGK	upstream gene variant	4564	ENST00000468601	?	?	1
NAGK	upstream gene variant	2685	ENST00000524537	?	ENSP00000437192	1
NAGK	downstream gene variant	2471	ENST00000531799	?	?	1
NAGK	upstream gene variant	3768	ENST00000490998	?	?	1
NAGK	upstream gene variant	697	ENST00000479854	?	?	1
RP11-467P9.1	downstream gene variant, upstream gene variant	2127	ENST00000608897	?	?	2

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
NAGK	ENST00000464638	?	?	1
NAGK	ENST00000455197	?	ENSP00000408550	1
NAGK	ENST00000524736	?	ENSP00000435437	1
NAGK	ENST00000613852	NM_017567.4	ENSP00000477639	1
NAGK	ENST00000533981	?	ENSP00000435207	1
NAGK	ENST00000493102	?	?	1
NAGK	ENST00000443938	?	ENSP00000411846	1
NAGK	ENST00000484984	?	?	1
NAGK	ENST00000531799	?	?	1
NAGK	ENST00000450272	?	ENSP00000400941	1
NAGK	ENST00000475709	?	?	1
NAGK	ENST00000418807	?	ENSP00000396070	1
NAGK	ENST00000428360	?	?	1
NAGK	ENST00000244204	?	ENSP00000244204	1
NAGK	ENST00000531934	?	ENSP00000436326	1
NAGK	ENST00000472519	?	?	1
NAGK	ENST00000455662	?	ENSP00000389087	1
NAGK	ENST00000443872	?	ENSP00000404531	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
NAGK	ENST00000480411	?	1

NAGK	ENST00000480411	?	1
NAGK	ENST00000465105	?	1
NAGK	ENST00000489309	?	1