Locus 86 (rs2000999)

SNiPAcard

Block annotations

Block info

genomic range	chr16:72,075,593-72,148,419
block size	72,827 bp
variant count	6 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.185 [-0.934 – 0.628]	**gene(s) hit or close-by**	DHX38, HP, HPR, PMFBP1, TXNL4B
phastCons	μ = 0.025 [0 – 0.128]	**eQTL gene(s)**	DHODH, DHX38, HP, HPR
GERP++	μ = 0.144 [-0.871 – 0.901]	**potentially regulated gene(s)**	–
CADD score	μ = 2.469 [0.059 – 6.223]	**disease gene(s)**	DHODH, HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Cholesterol, total	$<7.00\times10^{-41}$	GWAS Catalog	24097068	1
LDL cholesterol	$<4.00\times10^{-41}$	GWAS Catalog	24097068	1
Haptoglobin levels	$<8.00\times10^{-59}$	GWAS Catalog	22403646	1
Cholesterol, total	$<3.00\times10^{-24}$	GWAS Catalog	20686565	1
LDL cholesterol	$<2.00\times10^{-22}$	GWAS Catalog	20686565	1

Disease gene annotation

gene	trait	source DB	source entry/link	
DHODH	postaxial acrofacial dysostosis (POADS)	DECIPHER	MIM:263750	OMIM
DHODH	POSTAXIAL ACROFACIAL DYSOSTOSIS	OMIM	MIM:263750	OMIM
HP	ANHAPTOGLOBINEMIA	OMIM	MIM:614081	OMIM
DHODH	Postaxial acrofacial dysostosis	OrphaNet	OrphaNet:246	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DHODH	?	ENSG00000102967	muscularis mucosae	2.49×10^{-7} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	atrial appendage	9.97×10^{-11} (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	tibial artery	9.90×10^{-6} (p-value)	GTEx Portal V6	5
HP	?	ENSG00000257017	blood	1.01×10^{-9} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	blood	4.09×10^{-9} (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	putamen	5.64×10^{-11} (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	left ventricle	1.40×10^{-8} (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	subcutaneous adipocytes	9.37×10^{-6} (p-value)	GTEx Portal V6	4
HPR	?	ENSG00000261701	caudate basal ganglia	2.38×10^{-11} (p-value)	GTEx Portal V6	6

HP	?	ENSG00000257017	caudate basal ganglia	1.00×10-9 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cortex	3.50×10-17 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	nucleus accumbens	1.10×10-11 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	nucleus accumbens	2.56×10-6 (p-value)	GTEx Portal V6	1
DHODH	?	ENSG00000102967	tibial nerve	5.77×10-6 (p-value)	GTEx Portal V6	5
HPR	?	ENSG00000261701	anterior cingulate cortex	2.25×10-12 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	anterior cingulate cortex	2.94×10-8 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cerebellar hemisphere	3.70×10-15 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	cerebellum	3.66×10-23 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	hypothalamus	2.11×10-11 (p-value)	GTEx Portal V6	6
HPR	?	ENSG00000261701	frontal cortex	9.70×10-12 (p-value)	GTEx Portal V6	6
HP	?	ENSG00000257017	frontal cortex	9.37×10-6 (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	hippocampus	6.43×10-11 (p-value)	GTEx Portal V6	6
DHX38	?	ENSG00000140829	blood	2.46×10-3 (q-value)	SeeQTL DB (HapMap)	1

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	1612	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	1608	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	upstream gene variant, downstream gene variant	1611	ENST00000567142	?	ENSP00000454521	2
DHX38	upstream gene variant, downstream gene variant	1642	ENST00000563819	?	ENSP00000462397	2
DHX38	upstream gene variant	1643	ENST00000564622	?	?	1
DHX38	downstream gene variant	2100	ENST00000569935	?	?	1
DHX38	downstream gene variant	2733	ENST00000566329	?	?	1
DHX38	upstream gene variant, downstream gene variant	1976	ENST00000569952	?	?	2
DHX38	downstream gene variant	1617	ENST00000579387	?	ENSP00000462149	1
DHX38	upstream gene variant, downstream gene variant	1700	ENST00000567552	?	?	2
HP	downstream gene variant	2873	ENST00000562526	?	ENSP00000454413	1
HP	downstream gene variant	2919	ENST00000565574	?	ENSP00000454966	1
HP	downstream gene variant	3038	ENST00000228226	?	ENSP00000228226	1
HP	downstream gene variant	2874	ENST00000398131	NM_001126102.1	ENSP00000381199	1
HP	downstream gene variant	4110	ENST00000576168	?	ENSP00000461999	1
HP	downstream gene variant	2912	ENST00000357763	?	ENSP00000350406	1
HP	downstream gene variant	4740	ENST00000568417	?	ENSP00000462662	1
HP	downstream gene variant	4467	ENST00000569639	?	ENSP00000463755	1
HP	downstream gene variant	2874	ENST00000613898	?	ENSP00000478279	1
HP	downstream gene variant	2874	ENST00000566821	?	?	1

HP	downstream gene variant	2874	ENST00000566821	?	?	1
HP	downstream gene variant	3000	ENST00000567185	?	ENSP00000464070	1
HP	downstream gene variant	2873	ENST00000570083	?	ENSP00000457629	1
HP	downstream gene variant	2874	ENST00000355906	NM_005143.3	ENSP00000348170	1
HP	downstream gene variant	2902	ENST00000567612	?	ENSP00000463491	1
HP	downstream gene variant	3294	ENST00000564499	?	ENSP00000456503	1
PMFBP1	downstream gene variant	4577	ENST00000537465	?	ENSP00000443817	1
PMFBP1	downstream gene variant	4580	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	downstream gene variant	4581	ENST00000379073	?	ENSP00000368364	1
PMFBP1	downstream gene variant	4577	ENST00000237353	NM_031293.2	ENSP00000237353	1
TXNL4B	downstream gene variant	2595	ENST00000562153	?	ENSP00000454635	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DHX38	ENST00000562774	?	ENSP00000462965	1
DHX38	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	ENST00000579387	?	ENSP00000462149	1
HPR	ENST00000566168	?	?	3
HPR	ENST00000561690	?	ENSP00000462916	3
HPR	ENST00000356967	?	ENSP00000349451	3
HPR	ENST00000540303	NM_020995.3	ENSP00000441828	3
PMFBP1	ENST00000537792	?	ENSP00000443366	1
TXNL4B	ENST00000562153	?	ENSP00000454635	3