SNiPAcard

Block annotations

Block info

genomic range	chr16:72,078,043-72,227,398
block size	149,356 bp
variant count	7 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.615 [-2.712 – 0.203]	**gene(s) hit or close-by**	DHX38, PMFBP1, TXNL4B
phastCons	μ = 0.119 [0 – 0.83]	**eQTL gene(s)**	DHODH, DHX38, HP, HPR, TAT-AS1
GERP++	μ = -1.104 [-3.54 – 0.149]	**potentially regulated gene(s)**	–
CADD score	μ = 4.966 [0.747 – 11.04]	**disease gene(s)**	DHODH, HP

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
gamma-glutamyltyrosine	9.15×10^{-5}	Metabolomics GWAS Server	24816252	1

Disease gene annotation

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

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
TAT-AS1	?	ENSG00000260886	testis	2.38×10^{-6} (p-value)	GTEx Portal V6	4
DHODH	?	ENSG00000102967	tibial nerve	1.02×10^{-7} (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	transformed fibroblasts	3.63×10^{-6} (p-value)	GTEx Portal V6	5
DHODH	?	ENSG00000102967	tibial artery	2.90×10^{-6} (p-value)	GTEx Portal V6	3
HPR	?	ENSG00000261701	left ventricle	4.07×10^{-7} (p-value)	GTEx Portal V6	4
HPR	?	ENSG00000261701	blood	4.70×10^{-15} (p-value)	GTEx Portal V6	7
DHODH	?	ENSG00000102967	muscularis mucosae	1.53×10^{-5} (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	nucleus accumbens	5.64×10^{-6} (p-value)	GTEx Portal V6	1
DHX38	?	ENSG00000140829	EBV lymphocytes	3.30×10^{-6} (p-value)	GTEx Portal V6	1
HP	ENST00000570083	ILMN_1812433	monocyte	1.33×10^{-10} (p-value)	Fairfax et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					

HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000567612					
HP	ENST00000355906					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					
HP	ENST00000570083	ILMN_1812433	monocyte	4.52×10-48 (p-value)	Zeller et al.	1
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					
HP	ENST00000564499					
HP	ENST00000357763					

Putative effect on regulation

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	1318	ENST00000566794	?	ENSP00000455939	1
DHX38	downstream gene variant	1397	ENST00000564622	?	?	1
DHX38	downstream gene variant	3112	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	downstream gene variant	3116	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	1363	ENST00000566489	?	ENSP00000457887	1
DHX38	upstream gene variant	2852	ENST00000566329	?	?	1
DHX38	upstream gene variant	4871	ENST00000569935	?	?	1
DHX38	downstream gene variant	3385	ENST00000563819	?	ENSP00000462397	1
DHX38	downstream gene variant	3115	ENST00000567142	?	ENSP00000454521	1
DHX38	downstream gene variant	3374	ENST00000567552	?	?	1
DHX38	downstream gene variant	519	ENST00000569952	?	?	1
DHX38	downstream gene variant	3121	ENST00000579387	?	ENSP00000462149	1
PMFBP1	downstream gene variant	1882	ENST00000537792	?	ENSP00000443366	1
PMFBP1	downstream gene variant	3120	ENST00000535461	?	ENSP00000444953	1
PMFBP1	downstream gene variant	3077	ENST00000379073	?	ENSP00000368364	1
PMFBP1	downstream gene variant	2153	ENST00000537392	?	ENSP00000439564	1
PMFBP1	downstream gene variant	3073	ENST00000237353	NM_031293.2	ENSP00000237353	1
PMFBP1	downstream gene variant	3076	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	downstream gene variant	3073	ENST00000537465	?	ENSP00000443817	1
TXNL4B	upstream gene variant	3799	ENST00000426362	NM_001142318.1	ENSP00000392310	1
TXNL4B	upstream gene variant	4361	ENST00000569767	?	ENSP00000457699	1
TXNL4B	upstream gene variant	4579	ENST00000268483	NM_017853.2	ENSP00000268483	1
TXNL4B	downstream gene variant, upstream gene variant	145	ENST00000562153	?	ENSP00000454635	2

TXNL4B	downstream gene variant, upstream gene variant	145	ENST00000562153	?	ENSP00000454635	2
TXNL4B	upstream gene variant	4361	ENST00000423037	NM_001142317.1	ENSP00000408130	1
TXNL4B	upstream gene variant	4678	ENST00000565171	?	ENSP00000455550	1

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
DHX38	ENST00000567142	?	ENSP00000454521	1
DHX38	ENST00000563650	?	?	1
DHX38	ENST00000562774	?	ENSP00000462965	1
DHX38	ENST00000564307	?	?	1
DHX38	ENST00000567552	?	?	1
DHX38	ENST00000579387	?	ENSP00000462149	2
DHX38	ENST00000268482	NM_014003.3	ENSP00000268482	2
DHX38	ENST00000563819	?	ENSP00000462397	1
PMFBP1	ENST00000537792	?	ENSP00000443366	2
PMFBP1	ENST00000379073	?	ENSP00000368364	2
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	2
PMFBP1	ENST00000537465	?	ENSP00000443817	2
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	2
PMFBP1	ENST00000537392	?	ENSP00000439564	1