SNiPAcard

Block annotations

Block info

genomic range	chr16:72,151,101-72,243,277
block size	92,177 bp
variant count	37 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = 0.280 [-1.415 – 3.761]	**gene(s) hit or close-by**	DHX38, PMFBP1
phastCons	μ = 0.151 [0 – 1]	**eQTL gene(s)**	DHODH, DHX38, HP, HPR, PMFBP1, TXNL4B
GERP++	μ = 0.419 [-6.41 – 6.17]	**potentially regulated gene(s)**	PMFBP1
CADD score	μ = 5.929 [0.052 – 27.3]	**disease gene(s)**	DHODH, HP

Trait annotations

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 transcript

Amino acid sequence alteration

gene	effect type	affected transcript	RefSeq id	protein	exchanged AA's	exchanged codons	SIFT prediction	PolyPhen prediction	variant(s)
PMFBP1	missense variant (splice region), missense variant	ENST00000537465	?	ENSP00000443817	2	2			2
PMFBP1	missense variant (splice region), missense variant	ENST00000355636	NM_001160213.1	ENSP00000347854	2	2			2
PMFBP1	missense variant (splice region), missense variant	ENST00000237353	NM_031293.2	ENSP00000237353	2	2			2

Amino acid sequence alteration (splice region)

gene	effect type	affected transcript	RefSeq id	protein	amino acid	codon	SIFT prediction	PolyPhen prediction	varian(s)
PMFBP1	missense variant	ENST00000537392	?	ENSP00000439564	K/T	aAg/aCg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
DHODH	?	ENSG00000102967	muscularis mucosae	1.31×10-7 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	tibial nerve	8.46×10-12 (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	tibial nerve	3.29×10-7 (p-value)	GTEx Portal V6	27
HP	?	ENSG00000257017	tibial nerve	4.59×10-10 (p-value)	GTEx Portal V6	6
DHODH	?	ENSG00000102967	transformed fibroblasts	1.15×10-9 (p-value)	GTEx Portal V6	37

DHODH	?	ENSG00000102967	transformed fibroblasts	1.15×10-9 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	tibial artery	3.19×10-12 (p-value)	GTEx Portal V6	18
DHODH	?	ENSG00000102967	tibial artery	5.31×10-8 (p-value)	GTEx Portal V6	30
HP	?	ENSG00000257017	tibial artery	7.12×10-7 (p-value)	GTEx Portal V6	5
TXNL4B	?	ENSG00000140830	tibial artery	8.53×10-7 (p-value)	GTEx Portal V6	3
PMFBP1	?	ENSG00000118557	esophagus mucosa	9.96×10-8 (p-value)	GTEx Portal V6	33
DHODH	?	ENSG00000102967	skeletal muscle	3.84×10-6 (p-value)	GTEx Portal V6	23
DHODH	?	ENSG00000102967	transverse colon	3.61×10-8 (p-value)	GTEx Portal V6	35
HPR	?	ENSG00000261701	aorta	4.73×10-12 (p-value)	GTEx Portal V6	37
DHODH	?	ENSG00000102967	subcutaneous adipocytes	7.14×10-9 (p-value)	GTEx Portal V6	37
HPR	?	ENSG00000261701	subcutaneous adipocytes	3.60×10-9 (p-value)	GTEx Portal V6	5
PMFBP1	ENST00000355636	ILMN_1713867	adipocyte	1.78×10-5 (p-value)	MuTHER consortium	7
PMFBP1	ENST00000237353					
PMFBP1	ENST00000537465					
PMFBP1	ENST00000379073					
DHX38	ENST00000567552	ILMN_1735679	blood	9.66×10-11 (p-value)	MuTHER consortium	10
DHX38	ENST00000268482					
DHX38	ENST00000563819					
DHX38	ENST00000579387					
HP	ENST00000570083	ILMN_1812433	monocyte	2.30×10-11 (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	?	ENSG00000257017	blood	2.98×10-6 (p-value)	GTEx Portal V6	2
HPR	?	ENSG00000261701	sun exposed skin	2.75×10-7 (p-value)	GTEx Portal V6	5
DHODH	?	ENSG00000102967	sun exposed skin	1.39×10-5 (p-value)	GTEx Portal V6	1
PMFBP1	ENST00000355636	ILMN_1713867	monocyte	4.80×10-13 (p-value)	Zeller et al.	4
PMFBP1	ENST00000237353					
PMFBP1	ENST00000537465					
PMFBP1	ENST00000379073					
HP	ENST00000570083	ILMN_1812433	monocyte	1.16×10-40 (p-value)	Zeller et al.	4
HP	ENST00000567185					
HP	ENST00000566821					
HP	ENST00000613898					
HP	ENST00000565574					
HP	ENST00000228226					
HP	ENST00000355906					
HP	ENST00000567612					
HP	ENST00000398131					

HP	ENST00000564499					
HP	ENST00000357763					
HPR	?	ENSG00000261701	thyroid	2.10×10-7 (p-value)	GTEx Portal V6	3
DHX38	ENST00000567552	ILMN_1735679	blood	6.16×10-7 (p-value)	Westra et al.	2
DHX38	ENST00000268482					
DHX38	ENST00000563819					
DHX38	ENST00000579387					
PMFBP1	?	ENSG00000118557	EBV lymphocytes	8.30×10-6 (p-value)	GTEx Portal V6	1
HPR	?	ENSG00000261701	breast	4.30×10-6 (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000021719		

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000053177 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF, USF1, DNase1
		HSMMtube	DNase1, CTCF
		blood (K562)	Rad21, CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	DNase1, Rad21, USF1, H3K4me1, CTCF
		blood (GM12878)	Rad21, CTCF
		lung (IMR90)	CTCF
		nervous (NH-A)	DNase1, CTCF
		skin (NHEK)	CTCF
		NHLF	CTCF
		Osteobl	H2AZ, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF, DNase1
		monocytes (Monocytes-CD14+)	CTCF, H3K4me1
		endothelium (HUVEC)	CTCF
		A549	CTCF
ENSR00000509744 (promoter flanking region)	2	cervix (HeLa-S3)	DNase1
		NHLF	DNase1
		monocytes (Monocytes-CD14+)	DNase1
		endothelium (HUVEC)	Cjun
		liver (HepG2)	DNase1
		Osteobl	H3K9me3, H3K27ac
		skin (NHDF-AD)	DNase1
ENSR00000665811 (CTCF binding site)	1	cervix (HeLa-S3)	CTCF
		endothelium (HUVEC)	CTCF
		embryonic stem cell (H1ESC)	Rad21, CTCF, DNase1
		liver (HepG2)	Rad21, CTCF
		blood (K562)	CTCF, Rad21, H3K27me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
DHX38	downstream gene variant	4299	ENST00000579387	?	ENSP00000462149	1
DHX38	downstream gene variant	4294	ENST00000562774	?	ENSP00000462965	1
DHX38	downstream gene variant	4293	ENST00000567142	?	ENSP00000454521	1

DHX38	downstream gene variant	4290	ENST00000268482	NM_014003.3	ENSP00000268482	1
DHX38	downstream gene variant	4563	ENST00000563819	?	ENSP00000462397	1
DHX38	downstream gene variant	4552	ENST00000567552	?	?	1
PMFBP1	downstream gene variant, upstream gene variant	344	ENST00000237353	NM_031293.2	ENSP00000237353	4
PMFBP1	upstream gene variant	4214	ENST00000537792	?	ENSP00000443366	1
PMFBP1	downstream gene variant, upstream gene variant	363	ENST00000536211	?	ENSP00000439841	4
PMFBP1	downstream gene variant, upstream gene variant	344	ENST00000537465	?	ENSP00000443817	4
PMFBP1	downstream gene variant, upstream gene variant	347	ENST00000355636	NM_001160213.1	ENSP00000347854	3
PMFBP1	downstream gene variant, upstream gene variant	348	ENST00000379073	?	ENSP00000368364	4
PMFBP1	downstream gene variant, upstream gene variant	107	ENST00000535461	?	ENSP00000444953	3
PMFBP1	downstream gene variant, upstream gene variant	236	ENST00000539172	?	ENSP00000440941	3
PMFBP1	downstream gene variant, upstream gene variant	249	ENST00000543746	?	?	4
PMFBP1	downstream gene variant, upstream gene variant	15	ENST00000537392	?	ENSP00000439564	7
PMFBP1	downstream gene variant, upstream gene variant	43	ENST00000540440	?	ENSP00000440755	7

Putative effect on transcript

3'-UTR variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000379073	?	ENSP00000368364	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000536211	?	ENSP00000439841	11
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	22
PMFBP1	ENST00000537792	?	ENSP00000443366	7
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	22
PMFBP1	ENST00000543746	?	?	11
PMFBP1	ENST00000537465	?	ENSP00000443817	22
PMFBP1	ENST00000535461	?	ENSP00000444953	12
PMFBP1	ENST00000379073	?	ENSP00000368364	22
PMFBP1	ENST00000539172	?	ENSP00000440941	11
PMFBP1	ENST00000537392	?	ENSP00000439564	5
PMFBP1	ENST00000540440	?	ENSP00000440755	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
PMFBP1	ENST00000355636	NM_001160213.1	ENSP00000347854	1
PMFBP1	ENST00000237353	NM_031293.2	ENSP00000237353	1
PMFBP1	ENST00000537465	?	ENSP00000443817	1
PMFBP1	ENST00000379073	?	ENSP00000368364	2