SNiPAcard

Block annotations

Block info

genomic range	chr11:18,435,241-18,474,128
block size	38,888 bp
variant count	94 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.590 [-3.912 – 2.718]	**gene(s) hit or close-by**	LDHAL6A, LDHC, RP11-613F22.7
phastCons	μ = 0.112 [0 – 0.908]	**eQTL gene(s)**	LDHAL6A, YWHABP2
GERP++	μ = -0.091 [-4.48 – 2.45]	**potentially regulated gene(s)**	AC084117.3, GTF2H1, LDHA, LDHC, RP11-113D6.3, RP11-137N23.1, RP11-613F22.6, SERGEF, SPTY2D1, TSG101, UEVLD
CADD score	μ = 4.204 [0.024 – 15.14]	**disease gene(s)**	LDHA

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Amyotrophic lateral sclerosis (sporadic)	<9.00×10-6	GWAS Catalog	24529757	1

Disease gene annotation

gene	trait	source DB	source entry/link
LDHA	GLYCOGEN STORAGE DISEASE XI	OMIM	MIM:612933
LDHA	Glycogen storage disease due to lactate dehydrogenase M-subunit deficiency	OrphaNet	OrphaNet:284426

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
YWHABP2	?	ENSG00000256464	skeletal muscle	5.30×10-8 (p-value)	GTEx Portal V6	94
LDHAL6A	?	ENSG00000166800	muscularis mucosae	5.05×10-6 (p-value)	GTEx Portal V6	71
LDHAL6A	?	ENSG00000166800	pituitary	2.69×10-7 (p-value)	GTEx Portal V6	6

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000099956	1	ENCP00000011421	LDHC
		ENCP00000011426	TSG101 RP11-613F22.6
		ENCP00000011382	SERGEF
		ENCP00000011436	RP11-137N23.1
		ENCP00000011396	RP11-113D6.3
		ENCP00000011430	UEVLD
		ENCP00000011429	UEVLD

ENCE00000100103	1	ENCP00000011410	GTF2H1
		ENCP00000011435	SPTY2D1
		ENCP00000011419	LDHA
		ENCP00000011420	AC084117.3
		ENCP00000011418	LDHA

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
LDHAL6A	upstream gene variant	3303	ENST00000280706	NM_144972.4	ENSP00000280706	3
LDHAL6A	upstream gene variant	3243	ENST00000396213	NM_001144071.1	ENSP00000379516	3
LDHAL6A	upstream gene variant	3243	ENST00000615355	?	ENSP00000478764	3
LDHC	downstream gene variant	756	ENST00000396215	?	ENSP00000379518	3
LDHC	downstream gene variant	872	ENST00000535809	?	ENSP00000443997	3
LDHC	downstream gene variant	872	ENST00000546146	?	ENSP00000443414	3
LDHC	downstream gene variant	756	ENST00000545848	?	ENSP00000438196	3
LDHC	downstream gene variant	872	ENST00000536880	?	ENSP00000439555	3
LDHC	downstream gene variant	756	ENST00000280704	NM_017448.3, NM_002301.4	ENSP00000280704	3
LDHC	downstream gene variant	872	ENST00000537486	?	ENSP00000441478	3
LDHC	downstream gene variant	872	ENST00000544105	?	ENSP00000439060	3
LDHC	downstream gene variant	756	ENST00000539369	?	ENSP00000441991	3
LDHC	downstream gene variant	523	ENST00000541669	?	ENSP00000437783	1
RP11-613F22.7	downstream gene variant, upstream gene variant	32	ENST00000535082	?	?	14

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
LDHC	ENST00000396215	?	ENSP00000379518	91
LDHC	ENST00000535809	?	ENSP00000443997	91
LDHC	ENST00000280704	NM_017448.3, NM_002301.4	ENSP00000280704	91
LDHC	ENST00000541669	?	ENSP00000437783	91
LDHC	ENST00000545848	?	ENSP00000438196	91
LDHC	ENST00000536880	?	ENSP00000439555	91
LDHC	ENST00000546146	?	ENSP00000443414	91
LDHC	ENST00000537486	?	ENSP00000441478	91
LDHC	ENST00000544105	?	ENSP00000439060	91
LDHC	ENST00000539369	?	ENSP00000441991	91

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
LDHC	ENST00000541669	?	ENSP00000437783	2