SNiPAcard

Block annotations

Block info

genomic range	chr14:74,475,796-74,661,497
block size	185,702 bp
variant count	59 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-3.067 – 2.691]	**gene(s) hit or close-by**	ALDH6A1, CCDC176, ENTPD5, LIN52, RN7SL530P, RP5-892G5.2
phastCons	μ = 0.080 [0 – 1]	**eQTL gene(s)**	ALDH6A1, COQ6, LIN52, PTGR2, RP5-1021I20.2
GERP++	μ = -0.003 [-7.89 – 5.78]	**potentially regulated gene(s)**	–
CADD score	μ = 4.529 [0.029 – 17.19]	**disease gene(s)**	COQ6, ALDH6A1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
COQ6	COENZYME Q10 DEFICIENCY, PRIMARY, 6	OMIM	MIM:614650
ALDH6A1	METHYLMALONATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	OMIM	MIM:614105
COQ6	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	OrphaNet	OrphaNet:280406
ALDH6A1	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	OrphaNet	OrphaNet:289307

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)
CCDC176	missense variant	ENST00000464394	?	ENSP00000451659	D/V	gAt/gTt	tolerated	benign	1
CCDC176	missense variant	ENST00000394009	NM_025057.2	ENSP00000377577	D/V	gAt/gTt	tolerated	benign	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
COQ6	ENST00000554320	ILMN_1783985	skin	7.62×10^{-7} (p-value)	MuTHER consortium	20
COQ6	ENST00000394026		blood	3.46×10^{-6} (p-value)	MuTHER consortium	20
COQ6	ENST00000556299					
COQ6	ENST00000629426					
COQ6	ENST00000555511					
COQ6	ENST00000238709					
COQ6	ENST00000334571					
COQ6	ENST00000556588					
COQ6	ENST00000554341					
COQ6	ENST00000554920	ILMN_1664367	blood	1.25×10^{-5} (p-value)	MuTHER consortium	19
COQ6	ENST00000554193					
COQ6	ENST00000553922					
COQ6	ENST00000555552					

COQ6	ENST00000394026					
COQ6	ENST00000555511					
COQ6	ENST00000334571					
COQ6	ENST00000238709					
COQ6	ENST00000556300					
COQ6	ENST00000555196					
LIN52	ENST00000554076	ILMN_1797055	skin	1.75×10-12 (p-value)	MuTHER consortium	20
LIN52	ENST00000555028		blood	1.95×10-9 (p-value)	MuTHER consortium	20
			adipocyte	5.39×10-10 (p-value)	MuTHER consortium	20
LIN52	?	ENSG00000205659	pancreas	1.25×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	lung	9.83×10-11 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	lung	1.06×10-7 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	atrial appendage	1.17×10-6 (p-value)	GTEx Portal V6	55
PTGR2	?	ENSG00000140043	atrial appendage	4.62×10-7 (p-value)	GTEx Portal V6	38
LIN52	?	ENSG00000205659	transformed fibroblasts	2.58×10-7 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	tibial artery	2.29×10-8 (p-value)	GTEx Portal V6	58
LIN52	?	ENSG00000205659	breast	2.75×10-7 (p-value)	GTEx Portal V6	57
COQ6	?	ENSG00000119723	thyroid	2.86×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	thyroid	2.43×10-12 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	thyroid	1.22×10-7 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	skeletal muscle	6.97×10-8 (p-value)	GTEx Portal V6	59
COQ6	?	ENSG00000119723	skeletal muscle	6.68×10-6 (p-value)	GTEx Portal V6	15
LIN52	?	ENSG00000205659	EBV lymphocytes	3.22×10-9 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	unexposed skin	1.03×10-6 (p-value)	GTEx Portal V6	58
LIN52	?	ENSG00000205659	sun exposed skin	3.00×10-11 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	aorta	1.11×10-10 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	aorta	1.23×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	subcutaneous adipocytes	2.30×10-14 (p-value)	GTEx Portal V6	59
RP5-1021I20.2	?	ENSG00000258586	tibial nerve	1.51×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	tibial nerve	8.71×10-16 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	esophagus mucosa	6.24×10-8 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	esophagus mucosa	7.60×10-6 (p-value)	GTEx Portal V6	4
LIN52	?	ENSG00000205659	adrenal gland	4.23×10-8 (p-value)	GTEx Portal V6	59
LIN52	?	ENSG00000205659	testis	1.87×10-11 (p-value)	GTEx Portal V6	59
PTGR2	?	ENSG00000140043	visceral adipocytes	7.09×10-6 (p-value)	GTEx Portal V6	2
LIN52	?	ENSG00000205659	muscularis mucosae	1.67×10-7 (p-value)	GTEx Portal V6	21
LIN52	?	ENSG00000205659	coronary artery	4.72×10-6 (p-value)	GTEx Portal V6	1

LIN52	ENST00000554076	ILMN_1797055	monocyte	2.78×10^{-9} (p-value)	Fairfax et al.	4
LIN52	ENST00000555028		b-cell	1.34×10^{-7} (p-value)	Fairfax et al.	4
ALDH6A1	ENST00000554501	ILMN_2096985	blood	5.18×10^{-5} (p-value)	Westra et al.	1
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00000420800 (TF binding site)	1	endothelium (HUVEC)	Cjun, DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00000098742 (CTCF binding site)	1	embryonic stem cell (H1ESC)	Rad21, CTCF
		HSMMtube	CTCF
		blood (K562)	CTCF
		skin (NHDF-AD)	CTCF
		muscle (HSMM)	CTCF
		liver (HepG2)	Rad21, CTCF
		lung (IMR90)	CTCF
		blood (GM12878)	Rad21, CTCF
		nervous (NH-A)	CTCF
		skin (NHEK)	DNase1
		NHLF	CTCF
		Osteobl	CTCF
		blood (DND-41)	CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	CTCF
		endothelium (HUVEC)	H3K36me3, CTCF
		A549	CTCF, DNase1
ENSR00000420807 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac
		cervix (HeLa-S3)	DNase1, TAF1, H3K27ac
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		skin (NHEK)	H3K27ac
ENSR00000420810 (enhancer)	1	endothelium (HUVEC)	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		liver (HepG2)	H3K4me1

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ALDH6A1	upstream gene variant	97	ENST00000554501	?	?	1
ALDH6A1	upstream gene variant	126	ENST00000553814	?	?	1
ALDH6A1	upstream gene variant	99	ENST00000350259	NM_001278593.1	ENSP00000342564	1
ALDH6A1	upstream gene variant	54	ENST00000554231	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	99	ENST00000556852	?	?	2

ALDH6A1	upstream gene variant	53	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	1
ALDH6A1	upstream gene variant	2347	ENST00000555126	?	ENSP00000452081	2
CCDC176	downstream gene variant	4691	ENST00000492247	?	?	1
CCDC176	upstream gene variant, downstream gene variant	1683	ENST00000492026	?	?	2
CCDC176	downstream gene variant	2456	ENST00000463558	?	ENSP00000480689	1
CCDC176	upstream gene variant	96	ENST00000477986	?	?	2
LIN52	upstream gene variant, downstream gene variant	959	ENST00000554289	?	?	4
LIN52	upstream gene variant	410	ENST00000554938	?	ENSP00000452513	3
LIN52	upstream gene variant, downstream gene variant	417	ENST00000554076	?	?	6
LIN52	upstream gene variant	414	ENST00000553404	?	?	3
LIN52	upstream gene variant	250	ENST00000555028	NM_001024674.2	ENSP00000451812	3
RN7SL530P	upstream gene variant, downstream gene variant	833	ENST00000485097	?	?	3
RP5-892G5.2	upstream gene variant, downstream gene variant	1971	ENST00000555526	?	?	3

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000553814	?	?	5
ALDH6A1	ENST00000556852	?	?	4
ALDH6A1	ENST00000554501	?	?	9
ALDH6A1	ENST00000554231	?	?	5
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	9
ALDH6A1	ENST00000555126	?	ENSP00000452081	4
ALDH6A1	ENST00000350259	NM_001278593.1	ENSP00000342564	9
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	12
CCDC176	ENST00000489323	?	?	8
CCDC176	ENST00000463558	?	ENSP00000480689	6
CCDC176	ENST00000492026	?	?	16
CCDC176	ENST00000464394	?	ENSP00000451659	8
ENTPD5	ENST00000556108	?	?	2
ENTPD5	ENST00000557325	?	ENSP00000451810	2
ENTPD5	ENST00000554664	?	?	2
ENTPD5	ENST00000557681	?	?	2
ENTPD5	ENST00000556242	?	ENSP00000451483	2
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	2
LIN52	ENST00000554938	?	ENSP00000452513	38
LIN52	ENST00000555028	NM_001024674.2	ENSP00000451812	38
LIN52	ENST00000554076	?	?	7
LIN52	ENST00000553404	?	?	38
LIN52	ENST00000554289	?	?	2

LIN52	ENST00000554289	?	?	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ALDH6A1	ENST00000554501	?	1
CCDC176	ENST00000489323	?	1
CCDC176	ENST00000492026	?	2
CCDC176	ENST00000477986	?	1
ENTPD5	ENST00000557681	?	1