Locus 61 (rs17094448)

SNiPAcard

Block annotations

Block info

genomic range	chr14:74,304,567-74,652,932
block size	348,366 bp
variant count	145 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.322 [-4.812 – 3.018]	**gene(s) hit or close-by**	AC005480.1, ALDH6A1, CCDC176, COQ6, ENTPD5, FAM161B, LIN52, PTGR2, RN7SL530P, RP5-1021I20.4, RP5-1021I20.5, RP5-1021I20.6, RP5-1021I20.7, RP5-1021I20.8, Y_RNA, Y_RNA, ZNF410
phastCons	μ = 0.075 [0 – 1]	**eQTL gene(s)**	ALDH6A1, CCDC176, ENTPD5, LIN52
GERP++	μ = -0.197 [-9.11 – 3.71]	**potentially regulated gene(s)**	ABCD4, PNMA1
CADD score	μ = 4.182 [0.01 – 21.5]	**disease gene(s)**	ALDH6A1, COQ6, ABCD4

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
ALDH6A1	METHYLMALONATE SEMIALDEHYDE DEHYDROGENASE DEFICIENCY	OMIM	MIM:614105
COQ6	COENZYME Q10 DEFICIENCY, PRIMARY, 6	OMIM	MIM:614650
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	OMIM	MIM:614857
ALDH6A1	Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency	OrphaNet	OrphaNet:289307
COQ6	Familial steroid-resistant nephrotic syndrome with sensorineural deafness	OrphaNet	OrphaNet:280406
ABCD4	Methylmalonic acidemia with homocystinuria, type cblJ	OrphaNet	OrphaNet:369955
ABCD4	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblJ TYPE	DECIPHER	MIM:614857

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ENTPD5	ENST00000334696	ILMN_1745849	blood	2.53×10^{-6} (p-value)	Westra et al.	24
LIN52	ENST00000554076	ILMN_1797055	blood	7.71×10^{-7} (p-value)	Westra et al.	10
LIN52	ENST00000555028					
CCDC176	ENST00000394009	ILMN_1806456	blood	4.46×10^{-5} (p-value)	Westra et al.	7
ALDH6A1	ENST00000554501	ILMN_2096985	b-cell	4.66×10^{-4} (p-value)	Fairfax et al.	1
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					
CCDC176	?	ENSG00000119636	blood	9.70×10^{-8} (p-value)	GTEx Portal V6	21
ALDH6A1	ENST00000554501	ILMN_2096985	monocyte	3.63×10^{-14} (p-value)	Zeller et al.	2
ALDH6A1	ENST00000350259					
ALDH6A1	ENST00000555126					
ALDH6A1	ENST00000553458					

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000171206	1	ENCP00000019795	PNMA1
		ENCP00000019810	ABCD4

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001623807 (CTCF binding site)	1	cervix (HeLa-S3)	DNase1
		embryonic stem cell (H1ESC)	DNase1
		blood (K562)	Egr1
ENSR00000652291 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, DNase1, H3K9ac, USF1, H3K27me3, CTCF, PolII, TAF7, H3K27ac, TAF1, H3K4me2
		HSMMtube	DNase1, H3K36me3, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	H3K4me3, DNase1, H3K4me2, PolII, TAF1, H2AZ, HEY1, Cmyc, H3K9ac, PU1, Max, H3K27ac
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, H3K4me2, H3K27ac
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	DNase1, PolII, TAF1, USF1, H3K4me2, H3K9ac, H3K27ac, Cmyc, H3K4me3, H3K27me3
		lung (IMR90)	DNase1, H4K8ac, H3K27ac, H3K18ac, H3K4me2, H4K5ac, H3K4me3, H3K9ac
		blood (GM12878)	H3K27ac, H3K4me2, H3K9ac, ZBTB33, H3K4me3, H2AZ, DNase1, TAF1, PolII
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	H3K27ac, H3K9ac, H3K4me2, DNase1, H3K4me3
		NHLF	H3K36me3, H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H3K4me3, H3K4me2, H3K27ac, H2AZ
		blood (DND-41)	H3K4me3, H3K9ac, H3K27ac, H3K4me1
		breast (HMEC)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, DNase1, Max, H3K4me3, H3K4me2, H3K9ac
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000420786 (promoter)	2	embryonic stem cell (H1ESC)	H3K36me3, PolII, Egr1, Gabp, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	H3K79me2, H3K9ac, H3K4me2, DNase1, H2AZ, H3K4me3, H3K27ac
		blood (K562)	H2AZ, H3K79me2, Cmyc, Gabp, H3K9ac, Max, H3K27ac, Egr1, TAF1, ELF1, PolII, H3K4me3, DNase1, H3K4me2
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2, H2AZ
		liver (HepG2)	DNase1, Gabp, PolII, H3K79me2, TAF1, ELF1, H2AZ, H3K4me2, H3K9ac, H3K27ac, H3K4me3
		lung (IMR90)	DNase1, H3K27ac, H3K18ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	PolII, H2AZ, DNase1, Egr1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K27ac, H3K9ac
		Osteobl	H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K27ac, H3K4me2, H3K4me1, H3K36me3, H3K4me3, H3K9ac
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, H3K79me2, Gabp, DNase1, PolII
		monocytes (Monocytes-CD14+)	DNase1, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K4me2, H3K4me3, H3K9ac, PolII, DNase1, Cmyc
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3
ENSR00000420795 (promoter)	2	embryonic stem cell (H1ESC)	Yy1, TAF1, H3K4me2, H3K4me3, DNase1, H3K9ac, Sin3Ak20, H3K27me3, CTCF, PolII, Rad21
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		blood (K562)	H2AZ, HEY1, Gabp, H3K9ac, H3K27ac, H3K4me3, DNase1, CTCF, H3K4me2, PolII
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, DNase1, H3K4me2, H3K4me1, Gabp, Yy1, PolII, TAF1, ELF1
		lung (IMR90)	CTCF, DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, Yy1, ELF1, H3K4me3, CTCF, H3K9ac, H2AZ, PolII
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, DNase1, H3K27ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac, CTCF
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2

		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K4me2, CTCF
		cervix (HeLa-S3)	DNase1, CTCF, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K27ac, H3K9ac, H3K4me3, H3K4me2, DNase1
		endothelium (HUVEC)	Cmyc, DNase1, Max, H3K4me3, H3K4me2, PolII
		A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00000652293 (promoter flanking region)	1	monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me3, PolII, H3K4me1, H3K4me2, H3K9ac, H3K27ac
		lung (IMR90)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00001623814 (enhancer)	2	liver (HepG2)	H3K4me1, H3K27ac
		lung (IMR90)	H3K36me3
		blood (DND-41)	H3K36me3
ENSR00000241136 (enhancer)	1	liver (HepG2)	H3K4me1
		blood (K562)	DNase1
ENSR00000180920 (promoter)	4	embryonic stem cell (H1ESC)	H3K9ac, H3K27me3, Sin3Ak20, H3K4me2, H3K4me3, DNase1
		HSMMtube	DNase1, H2AZ, H3K4me3, H3K4me2, H3K9ac
		blood (K562)	H3K27ac, Max, H3K4me3, DNase1, H3K4me2, PolII, H2AZ, Cmyc, H3K9ac, Srf
		skin (NHDF-AD)	H3K4me2, H3K9ac, DNase1, H3K4me3, H3K27ac
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	Srf, PolII, TAF1, H3K4me2, H3K9ac, H3K27ac, H3K4me3, DNase1
		lung (IMR90)	DNase1, H3K27ac, H3K4me2, H3K4me3, H3K9ac
		blood (GM12878)	DNase1, PolII, H2AZ, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	H3K27ac, H3K9ac, H3K4me3, DNase1
		Osteobl	H2AZ, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	PolII, H3K4me3, H3K27ac, H3K4me2, H3K9ac, Ini1, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, H3K9ac, H3K27ac, PolII, DNase1
		A549	H3K27ac, H3K9ac, H3K4me3, H3K4me2
ENSR00000420807 (promoter flanking region)	1	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
ENSR00000420808 (enhancer)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K4me1, H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	Cjun, H3K4me2
		liver (HepG2)	DNase1, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
ENSR00001623819 (CTCF binding site)	1	cervix (HeLa-S3)	H3K4me1, H3K4me2, DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K4me2
		embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	H3K36me3
ENSR00001623829 (CTCF binding site)	1	monocytes (Monocytes-CD14+)	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
AC005480.1	upstream gene variant, downstream gene variant	536	ENST00000624550	?	ENSP00000485283	20
ALDH6A1	downstream gene variant	267	ENST00000555126	?	ENSP00000452081	2
ALDH6A1	upstream gene variant	4119	ENST00000556852	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	2682	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	3
ALDH6A1	upstream gene variant	4146	ENST00000553814	?	?	1
ALDH6A1	downstream gene variant, upstream gene variant	247	ENST00000350259	NM_001278593.1	ENSP00000342564	3
ALDH6A1	downstream gene variant, upstream gene variant	4074	ENST00000554231	?	?	2
ALDH6A1	downstream gene variant, upstream gene variant	99	ENST00000554501	?	?	3
CCDC176	upstream gene variant, downstream gene variant	1365	ENST00000492247	?	?	4
CCDC176	upstream gene variant	133	ENST00000394009	NM_025057.2	ENSP00000377577	13
CCDC176	upstream gene variant	447	ENST00000492026	?	?	4
CCDC176	upstream gene variant	225	ENST00000463558	?	ENSP00000480689	12
CCDC176	upstream gene variant	223	ENST00000489323	?	?	12
CCDC176	upstream gene variant	130	ENST00000464394	?	ENSP00000451659	13
COQ6	upstream gene variant, downstream gene variant	308	ENST00000555392	?	?	5
COQ6	upstream gene variant, downstream gene variant	257	ENST00000553462	?	?	3
COQ6	upstream gene variant, downstream gene variant	1090	ENST00000554341	?	ENSP00000450736	2
COQ6	upstream gene variant, downstream gene variant	331	ENST00000334571	NM_182476.2	ENSP00000333946	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000554193	?	?	4
COQ6	upstream gene variant, downstream gene variant	337	ENST00000556300	?	?	3
COQ6	upstream gene variant, downstream gene variant	346	ENST00000554920	?	ENSP00000451562	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000553922	?	?	4
COQ6	upstream gene variant, downstream gene variant	348	ENST00000557205	?	?	3
COQ6	upstream gene variant, downstream gene variant	364	ENST00000238709	?	ENSP00000238709	3
COQ6	upstream gene variant, downstream gene variant	1076	ENST00000394026	NM_182480.2	ENSP00000377594	2
COQ6	upstream gene variant, downstream gene variant	1809	ENST00000553448	?	?	2
COQ6	upstream gene variant, downstream gene variant	1394	ENST00000556588	?	?	2
COQ6	upstream gene variant, downstream gene variant	339	ENST00000554153	?	ENSP00000451685	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000555552	?	?	4
COQ6	upstream gene variant, downstream gene variant	191	ENST00000557584	?	ENSP00000450511	3
COQ6	upstream gene variant, downstream gene variant	290	ENST00000629426	?	ENSP00000486650	3
COQ6	upstream gene variant, downstream gene variant	348	ENST00000554217	?	?	3
COQ6	upstream gene variant, downstream gene variant	346	ENST00000555196	?	ENSP00000451301	4
COQ6	upstream gene variant, downstream gene variant	1399	ENST00000556299	?	?	3
COQ6	upstream gene variant, downstream gene	562	ENST00000554320	?	ENSP00000451123	3

	variant					
COQ6	upstream gene variant, downstream gene variant	2347	ENST00000557780	?	?	2
COQ6	upstream gene variant, downstream gene variant	4160	ENST00000555511	?	?	2
ENTPD5	downstream gene variant, upstream gene variant	258	ENST00000555829	?	ENSP00000451499	4
ENTPD5	downstream gene variant, upstream gene variant	695	ENST00000554664	?	?	8
ENTPD5	downstream gene variant, upstream gene variant	432	ENST00000556242	?	ENSP00000451483	8
ENTPD5	downstream gene variant, upstream gene variant	352	ENST00000556108	?	?	9
ENTPD5	downstream gene variant, upstream gene variant	777	ENST00000557681	?	?	16
ENTPD5	downstream gene variant, upstream gene variant	549	ENST00000553284	?	ENSP00000451591	11
ENTPD5	downstream gene variant, upstream gene variant	762	ENST00000334696	NM_001249.2	ENSP00000335246	7
ENTPD5	downstream gene variant, upstream gene variant	317	ENST00000557325	?	ENSP00000451810	15
FAM161B	downstream gene variant, upstream gene variant	492	ENST00000286544	NM_152445.2	ENSP00000286544	6
FAM161B	downstream gene variant, upstream gene variant	174	ENST00000556794	?	ENSP00000450889	7
LIN52	downstream gene variant	46	ENST00000554289	?	?	2
PTGR2	upstream gene variant, downstream gene variant	413	ENST00000555661	?	ENSP00000452280	5
PTGR2	upstream gene variant, downstream gene variant	514	ENST00000267568	NM_152444.2	ENSP00000267568	6
PTGR2	upstream gene variant, downstream gene variant	1738	ENST00000553813	?	ENSP00000450824	4
PTGR2	upstream gene variant, downstream gene variant	438	ENST00000556219	?	?	5
PTGR2	upstream gene variant, downstream gene variant	411	ENST00000555228	NM_001146155.1, NM_001146154.1	ENSP00000450975	6
PTGR2	upstream gene variant, downstream gene variant	108	ENST00000555976	?	?	6
PTGR2	upstream gene variant, downstream gene variant	370	ENST00000553326	?	?	5
RN7SL530P	upstream gene variant	3981	ENST00000485097	?	?	1
RP5-1021I20.4	upstream gene variant, downstream gene variant	538	ENST00000556551	?	ENSP00000451484	4
RP5-1021I20.5	downstream gene variant	1666	ENST00000554009	?	?	1
RP5-1021I20.5	downstream gene variant, upstream gene variant	1565	ENST00000555916	?	?	3
RP5-1021I20.6	upstream gene variant, downstream gene variant	1270	ENST00000602874	?	?	5
RP5-1021I20.7	downstream gene variant, upstream gene variant	86	ENST00000604337	?	?	4
RP5-1021I20.8	downstream gene variant, upstream gene variant	762	ENST00000613926	?	?	8
Y_RNA	downstream gene variant, upstream gene variant	479	ENST00000362602	?	?	5
Y_RNA	downstream gene variant, upstream gene variant	361	ENST00000411368	?	?	5
ZNF410	upstream gene variant, downstream gene variant	488	ENST00000324593	NM_001242926.1	ENSP00000323293	6
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000442160	NM_001242924.1	ENSP00000407130	6
ZNF410	upstream gene variant, downstream gene variant	604	ENST00000554316	?	ENSP00000451748	6
ZNF410	upstream gene variant	544	ENST00000541357	?	?	5
ZNF410	upstream gene variant, downstream gene variant	342	ENST00000555620	?	?	5
ZNF410	upstream gene variant	1994	ENST00000557679	?	?	4
ZNF410	upstream gene variant, downstream gene variant	1176	ENST00000557214	?	ENSP00000451620	4
	upstream gene variant, downstream gene	542	ENST00000557363	?	ENSP00000452199	9

ZNF410	variant					
ZNF410	upstream gene variant, downstream gene variant	415	ENST00000555602	?	?	8
ZNF410	upstream gene variant, downstream gene variant	300	ENST00000540593	NM_001242927.1	ENSP00000442228	6
ZNF410	upstream gene variant	326	ENST00000556179	?	ENSP00000450884	3
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000412490	?	?	7
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000556659	?	ENSP00000452023	8
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000555044	NM_021188.2, NM_001242928.1	ENSP00000451763	6
ZNF410	upstream gene variant, downstream gene variant	2826	ENST00000615736	?	ENSP00000483073	6
ZNF410	upstream gene variant, downstream gene variant	1426	ENST00000554582	?	ENSP00000450650	8
ZNF410	upstream gene variant, downstream gene variant	1227	ENST00000334521	?	ENSP00000334170	3
ZNF410	upstream gene variant, downstream gene variant	554	ENST00000398139	?	ENSP00000381208	6
ZNF410	upstream gene variant, downstream gene variant	1157	ENST00000556797	?	ENSP00000451073	6
ZNF410	upstream gene variant, downstream gene variant	417	ENST00000555730	?	ENSP00000451807	9
ZNF410	upstream gene variant, downstream gene variant	2500	ENST00000554797	?	ENSP00000451116	5
ZNF410	upstream gene variant, downstream gene variant	741	ENST00000556160	?	ENSP00000451458	8
ZNF410	upstream gene variant, downstream gene variant	542	ENST00000557495	?	ENSP00000450569	8
ZNF410	upstream gene variant, downstream gene variant	491	ENST00000556396	?	ENSP00000451867	6

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
ENTPD5	ENST00000557325	?	ENSP00000451810	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000556852	?	?	2
ALDH6A1	ENST00000555126	?	ENSP00000452081	2
ALDH6A1	ENST00000554231	?	?	2
ALDH6A1	ENST00000553814	?	?	2
ALDH6A1	ENST00000350259	NM_001278593.1	ENSP00000342564	4
ALDH6A1	ENST00000554501	?	?	4
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	6
CCDC176	ENST00000464394	?	ENSP00000451659	12
CCDC176	ENST00000463558	?	ENSP00000480689	12
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	18
CCDC176	ENST00000492026	?	?	8
CCDC176	ENST00000489323	?	?	12
COQ6	ENST00000555552	?	?	1
COQ6	ENST00000553462	?	?	2
COQ6	ENST00000557205	?	?	2

COQ6	ENST00000555196	?	ENSP00000451301	1
COQ6	ENST00000553922	?	?	1
COQ6	ENST00000238709	?	ENSP00000238709	3
COQ6	ENST00000557584	?	ENSP00000450511	2
COQ6	ENST00000554153	?	ENSP00000451685	2
COQ6	ENST00000394026	NM_182480.2	ENSP00000377594	4
COQ6	ENST00000554341	?	ENSP00000450736	4
COQ6	ENST00000334571	NM_182476.2	ENSP00000333946	3
COQ6	ENST00000555511	?	?	2
COQ6	ENST00000557780	?	?	1
COQ6	ENST00000554320	?	ENSP00000451123	3
COQ6	ENST00000554193	?	?	1
COQ6	ENST00000554920	?	ENSP00000451562	3
COQ6	ENST00000556300	?	?	2
COQ6	ENST00000554217	?	?	1
COQ6	ENST00000629426	?	ENSP00000486650	3
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	55
ENTPD5	ENST00000557325	?	ENSP00000451810	46
ENTPD5	ENST00000555829	?	ENSP00000451499	1
ENTPD5	ENST00000553284	?	ENSP00000451591	12
ENTPD5	ENST00000554664	?	?	48
ENTPD5	ENST00000556242	?	ENSP00000451483	48
ENTPD5	ENST00000556108	?	?	40
ENTPD5	ENST00000557681	?	?	19
FAM161B	ENST00000286544	NM_152445.2	ENSP00000286544	5
FAM161B	ENST00000556794	?	ENSP00000450889	1
LIN52	ENST00000554289	?	?	2
LIN52	ENST00000553404	?	?	13
LIN52	ENST00000554938	?	ENSP00000452513	13
LIN52	ENST00000554076	?	?	5
LIN52	ENST00000555028	NM_001024674.2	ENSP00000451812	13
PTGR2	ENST00000553326	?	?	3
PTGR2	ENST00000555661	?	ENSP00000452280	17
PTGR2	ENST00000556219	?	?	14
PTGR2	ENST00000267568	NM_152444.2	ENSP00000267568	16
PTGR2	ENST00000555976	?	?	8
PTGR2	ENST00000555228	NM_001146155.1, NM_001146154.1	ENSP00000450975	16
PTGR2	ENST00000553813	?	ENSP00000450824	14
RP5-1021I20.4	ENST00000556551	?	ENSP00000451484	40
RP5-1021I20.5	ENST00000555916	?	?	13
RP5-1021I20.5	ENST00000554009	?	?	15

ZNF410	ENST00000334521	?	ENSP00000334170	21
ZNF410	ENST00000442160	NM_001242924.1	ENSP00000407130	23
ZNF410	ENST00000555044	NM_021188.2, NM_001242928.1	ENSP00000451763	23
ZNF410	ENST00000557363	?	ENSP00000452199	3
ZNF410	ENST00000557214	?	ENSP00000451620	7
ZNF410	ENST00000541357	?	?	18
ZNF410	ENST00000615736	?	ENSP00000483073	16
ZNF410	ENST00000556396	?	ENSP00000451867	23
ZNF410	ENST00000554797	?	ENSP00000451116	1
ZNF410	ENST00000540593	NM_001242927.1	ENSP00000442228	23
ZNF410	ENST00000556797	?	ENSP00000451073	1
ZNF410	ENST00000412490	?	?	10
ZNF410	ENST00000554316	?	ENSP00000451748	9
ZNF410	ENST00000555602	?	?	7
ZNF410	ENST00000555620	?	?	9
ZNF410	ENST00000555730	?	ENSP00000451807	3
ZNF410	ENST00000557679	?	?	11
ZNF410	ENST00000556659	?	ENSP00000452023	3
ZNF410	ENST00000398139	?	ENSP00000381208	23
ZNF410	ENST00000556179	?	ENSP00000450884	2
ZNF410	ENST00000554582	?	ENSP00000450650	7
ZNF410	ENST00000557495	?	ENSP00000450569	3
ZNF410	ENST00000556160	?	ENSP00000451458	3
ZNF410	ENST00000324593	NM_001242926.1	ENSP00000323293	23

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ALDH6A1	ENST00000553458	NM_005589.3, NM_001278594.1	ENSP00000450436	3
CCDC176	ENST00000394009	NM_025057.2	ENSP00000377577	3
ENTPD5	ENST00000334696	NM_001249.2	ENSP00000335246	1
FAM161B	ENST00000286544	NM_152445.2	ENSP00000286544	1
PTGR2	ENST00000555661	?	ENSP00000452280	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
COQ6	ENST00000394026	NM_182480.2	ENSP00000377594	1
COQ6	ENST00000554341	?	ENSP00000450736	1
ENTPD5	ENST00000553284	?	ENSP00000451591	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
COQ6	ENST00000553448	?	1
COQ6	ENST00000556588	?	1

ENTPD5	ENST00000554664	?	4
PTGR2	ENST00000556219	?	1
RP5-1021I20.5	ENST00000555916	?	2
RP5-1021I20.6	ENST00000602874	?	1