Locus 244 (rs1204656)

SNiPAcard

Block annotations

Block info

genomic range	chr20:33,887,955-34,023,219
block size	135,265 bp
variant count	39 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.104 [-1.732 – 4.902]	gene(s) hit or close-by	GDF5, GDF5OS, UQCC1
phastCons	μ = 0.086 [0 – 1]	eQTL gene(s)	CEP250, CPNE1, EDEM2, EIF6, ERGIC3, FAM83C, GDF5, GGT7, MMP24-AS1, PROCR, RPL36P4, UQCC1
GERP++	μ = -0.206 [-5.99 – 4.04]	potentially regulated gene(s)	ACSS2, CEP250, CPNE1, ERGIC3, FER1L4, GSS, MYH7B, NFS1, RBM12, ROMO1, TRPC4AP
CADD score	μ = 3.939 [0.19 – 20.8]	disease gene(s)	GDF5, MYH7B, NFS1, GSS

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
Height	<8.00×10-7	GWAS Catalog	18391951	1

Disease gene annotation

gene	trait	source DB	source entry/link
GDF5	Du Pan syndrome	DECIPHER	MIM:228900
GDF5	multiple synostoses syndrome type 2 (SYNS2)	DECIPHER	MIM:610017
GDF5	brachydactyly type C (BDC)	DECIPHER	MIM:113100
GDF5	brachydactyly type A2 (BDA2)	DECIPHER	MIM:112600
GDF5	brachydactyly type A1 (BDA1)	DECIPHER	MIM:112500
GDF5	symphalangism proximal syndrome (SYM1)	DECIPHER	MIM:185800
GDF5	acromesomelic chondrodysplasia Hunter-Thompson type (AMDH)	DECIPHER	MIM:201250
GDF5	acromesomelic chondrodysplasia Grebe type (AMDG)	DECIPHER	MIM:200700
GDF5	Acromesomelic dysplasia, Grebe type	OrphaNet	OrphaNet:2098
GDF5	Acromesomelic dysplasia, Hunter-Thomson type	OrphaNet	OrphaNet:968
GDF5	Proximal symphalangism	OrphaNet	OrphaNet:3250
GDF5	Multiple synostoses syndrome	OrphaNet	OrphaNet:3237
GDF5	Angel-shaped phalango-epiphyseal dysplasia	OrphaNet	OrphaNet:63442
GDF5	BRACHYDACTYLY TYPE C	OrphaNet	OrphaNet:93384
GDF5	Fibular aplasia - complex brachydactyly	OrphaNet	OrphaNet:2639
GDF5	BRACHYDACTYLY, TYPE A2	OrphaNet	OrphaNet:93396
GDF5	Brachydactyly, type A1	OrphaNet	OrphaNet:93388
MYH7B	Left ventricular noncompaction	OrphaNet	OrphaNet:54260
NFS1	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	OrphaNet	OrphaNet:397593
GSS	Glutathione synthetase deficiency with 5-oxoprolinuria	OrphaNet	OrphaNet:289846
GSS	Glutathione synthetase deficiency without 5-oxoprolinuria	OrphaNet	OrphaNet:289849
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY OF ERYTHROCYTES, HEMOLYTIC [...]	OMIM	MIM:231900
GSS	GLUTATHIONE SYNTHETASE DEFICIENCY	OMIM	MIM:266130

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
RPL36P4	?	ENSG00000224497	lung	9.15×10^{-9} (p-value)	GTEx Portal V6	38
UQCC1	?	ENSG00000101019	lung	1.53×10^{-7} (p-value)	GTEx Portal V6	39
GDF5	?	ENSG00000125965	lung	6.73×10^{-8} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	nucleus accumbens	5.91×10^{-7} (p-value)	GTEx Portal V6	28
RPL36P4	?	ENSG00000224497	tibial nerve	7.87×10^{-9} (p-value)	GTEx Portal V6	38
UQCC1	?	ENSG00000101019	tibial nerve	2.04×10^{-9} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	transformed fibroblasts	5.34×10^{-13} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	transformed fibroblasts	2.70×10^{-27} (p-value)	GTEx Portal V6	39
ERGIC3	?	ENSG00000125991	transformed fibroblasts	1.57×10^{-6} (p-value)	GTEx Portal V6	18
EIF6	?	ENSG00000242372	transformed fibroblasts	9.71×10^{-6} (p-value)	GTEx Portal V6	1
UQCC1	?	ENSG00000101019	blood	3.08×10^{-7} (p-value)	GTEx Portal V6	39
RPL36P4	?	ENSG00000224497	blood	1.32×10^{-5} (p-value)	GTEx Portal V6	2
UQCC1	?	ENSG00000101019	tibial artery	5.22×10^{-7} (p-value)	GTEx Portal V6	31
RPL36P4	?	ENSG00000224497	tibial artery	4.48×10^{-6} (p-value)	GTEx Portal V6	15
RPL36P4	?	ENSG00000224497	esophagus mucosa	3.72×10^{-9} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	esophagus mucosa	3.14×10^{-7} (p-value)	GTEx Portal V6	39
FAM83C	?	ENSG00000125998	esophagus mucosa	1.64×10^{-6} (p-value)	GTEx Portal V6	24
CEP250	?	ENSG00000126001	esophagus mucosa	9.34×10^{-6} (p-value)	GTEx Portal V6	9
RPL36P4	?	ENSG00000224497	thyroid	5.73×10^{-11} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	thyroid	2.91×10^{-7} (p-value)	GTEx Portal V6	37
CPNE1	?	ENSG00000214078	thyroid	1.35×10^{-5} (p-value)	GTEx Portal V6	1
RPL36P4	?	ENSG00000224497	skeletal muscle	9.81×10^{-7} (p-value)	GTEx Portal V6	32
UQCC1	?	ENSG00000101019	skeletal muscle	1.71×10^{-11} (p-value)	GTEx Portal V6	39
CEP250	?	ENSG00000126001	skeletal muscle	8.25×10^{-11} (p-value)	GTEx Portal V6	38
RPL36P4	?	ENSG00000224497	putamen	1.09×10^{-8} (p-value)	GTEx Portal V6	28
RPL36P4	?	ENSG00000224497	cerebellum	3.29×10^{-5} (p-value)	GTEx Portal V6	9
UQCC1	?	ENSG00000101019	sun exposed skin	5.91×10^{-10} (p-value)	GTEx Portal V6	39
EIF6	?	ENSG00000242372	sun exposed skin	8.34×10^{-6} (p-value)	GTEx Portal V6	6
RPL36P4	?	ENSG00000224497	sun exposed skin	1.18×10^{-6} (p-value)	GTEx Portal V6	31
RPL36P4	?	ENSG00000224497	subcutaneous adipocytes	3.54×10^{-11} (p-value)	GTEx Portal V6	39
UQCC1	?	ENSG00000101019	subcutaneous adipocytes	2.57×10^{-10} (p-value)	GTEx Portal V6	39
UQCC1	ENST00000374384	ILMN_2381121	monocyte	9.15×10^{-6} (p-value)	Fairfax et al.	2
UQCC1	ENST00000359226					
UQCC1	ENST00000457259					
UQCC1	ENST00000482440					
UQCC1	ENST00000374380					

Locus 244 (sentinel SNP rs1204656 + proxy variants in LD r2 > 0.8). SNiPA block annotation (www.snipa.org) based on 1000 Genomes Project phase 3 v5 (European population) variant set and Ensembl 82 annotations.

UQCC1	ENST00000472559					
UQCC1	ENST00000374394					
UQCC1	ENST00000453855					
UQCC1	ENST00000349714					
UQCC1	ENST00000496812					
UQCC1	ENST00000473982					
UQCC1	ENST00000424405					
UQCC1	ENST00000397556					
UQCC1	ENST00000374385					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	6.05×10-6 (p-value)	Fairfax et al.	2
CPNE1	ENST00000317677		b-cell	1.19×10-4 (p-value)	Fairfax et al.	2
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
GDF5	?	ENSG00000125965	pituitary	5.46×10-6 (p-value)	GTEx Portal V6	9
CPNE1	?	ENSG00000214078	blood	7.56×10-5 (q-value)	SeeQTL DB (HapMap)	6
RPL36P4	?	ENSG00000224497	adrenal gland	3.68×10-8 (p-value)	GTEx Portal V6	22
UQCC1	ENST00000374384	ILMN_2381121	blood	2.11×10-7 (p-value)	MuTHER consortium	12
UQCC1	ENST00000482440					
UQCC1	ENST00000457259					
UQCC1	ENST00000359226					
UQCC1	ENST00000374380					
UQCC1	ENST00000374394					
UQCC1	ENST00000472559					
UQCC1	ENST00000349714					
UQCC1	ENST00000453855					
UQCC1	ENST00000496812					
UQCC1	ENST00000473982					
UQCC1	ENST00000424405					
UQCC1	ENST00000397556					
UQCC1	ENST00000374385					
GGT7	ENST00000336431	ILMN_1737036	blood	5.13×10-5 (p-value)	MuTHER consortium	5
?	?	ILMN_1786186	skin	7.72×10-6 (p-value)	MuTHER consortium	7
CPNE1	ENST00000397443	ILMN_1670841	blood	1.49×10-9 (p-value)	MuTHER consortium	12
CPNE1	ENST00000317677		adipocyte	3.70×10-6 (p-value)	MuTHER consortium	12
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					

CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	blood	1.05×10^{-9} (p-value)	MuTHER consortium	12
CPNE1	ENST00000317677		adipocyte	8.95×10^{-8} (p-value)	MuTHER consortium	12
CPNE1	ENST00000498056		skin	2.73×10^{-5} (p-value)	MuTHER consortium	12
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
GDF5	?	ENSG00000125965	muscularis mucosae	1.09×10^{-5} (p-value)	GTEx Portal V6	7
RPL36P4	?	ENSG00000224497	muscularis mucosae	2.23×10^{-5} (p-value)	GTEx Portal V6	7
UQCC1	?	ENSG00000101019	visceral adipocytes	1.02×10^{-7} (p-value)	GTEx Portal V6	12
GDF5	?	ENSG00000125965	sigmoid colon	6.58×10^{-6} (p-value)	GTEx Portal V6	9
UQCC1	ENST00000374384	ILMN_1745152	monocyte	1.61×10^{-24} (p-value)	Zeller et al.	4
UQCC1	ENST00000457259					
UQCC1	ENST00000359226					
UQCC1	ENST00000374380					
UQCC1	ENST00000472559					
UQCC1	ENST00000374394					
UQCC1	ENST00000349714					
UQCC1	ENST00000496812					
UQCC1	ENST00000374385					
MMP24-AS1	ENST00000566203	ILMN_1911503	blood	3.89×10^{-6} (p-value)	Westra et al.	11
CEP250	ENST00000397527	ILMN_1799011	blood	1.12×10^{-6} (p-value)	Westra et al.	9
EDEM2	ENST00000374491	ILMN_1711909	blood	7.10×10^{-7} (p-value)	Westra et al.	2
EDEM2	ENST00000374492					
UQCC1	?	ENSG00000101019	unexposed skin	1.72×10^{-6} (p-value)	GTEx Portal V6	3
PROCR	?	ENSG00000101000	pancreas	4.03×10^{-6} (p-value)	GTEx Portal V6	5
RPL36P4	?	ENSG00000224497	frontal cortex	2.41×10^{-6} (p-value)	GTEx Portal V6	19
RPL36P4	?	ENSG00000224497	transverse colon	1.77×10^{-6} (p-value)	GTEx Portal V6	10
UQCC1	?	ENSG00000101019	transverse colon	4.32×10^{-6} (p-value)	GTEx Portal V6	2
RPL36P4	?	ENSG00000224497	testis	1.43×10^{-5} (p-value)	GTEx Portal V6	4
UQCC1	?	ENSG00000101019	breast	8.32×10^{-6} (p-value)	GTEx Portal V6	1

Putative effect on regulation

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000293024	1	ENCP00000031386	ERGIC3
		ENCP00000031342	GSS
		ENCP00000031336	ACSS2
		ENCP00000031392	FER1L4
		ENCP00000031410	NFS1 ROMO1
		ENCP00000031393	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031366	CEP250
		ENCP00000031346	MYH7B
		ENCP00000031405	CPNE1 RBM12
		ENCP00000031347	TRPC4AP

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001448537 (promoter flanking region)	4	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, Yy1, USF1, DNase1
		HSMMtube	CTCF, H3K4me1, H3K9ac, H3K27ac, DNase1
		blood (K562)	Cjun, Cfos, CTCF, H3K36me3, DNase1, HDAC2, USF1, Rad21, FOSL1, Max
		skin (NHDF-AD)	H3K27ac, DNase1, CTCF, H3K4me1
		muscle (HSMM)	H3K9ac, H3K27ac, CTCF, H3K4me1, DNase1, H3K36me3
		liver (HepG2)	FOSL2, Jund, Rad21, H3K4me1, H3K9ac, H3K27ac, CTCF, H3K36me3, DNase1
		blood (GM12878)	DNase1, Yy1, Rad21, CTCF, H3K36me3
		lung (IMR90)	H4K5ac, H3K36me3, H3K4ac, H2BK120ac, H2BK20ac, H2AK5ac, H4K91ac, CTCF, H3K18ac, H3K27ac, DNase1, H4K8ac, H2BK12ac
		nervous (NH-A)	DNase1, H3K9ac, CTCF, H3K27ac, H3K36me3
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K9ac, H3K27ac, H3K4me1
		NHLF	H3K36me3, H3K27ac, CTCF, DNase1, H3K4me1
		Osteobl	H3K27ac, H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K36me3, H3K4me1, H3K9ac, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, Gabp, Cmyc, Max, TAF1, H3K4me1, H3K27ac, Jund, PolII, BAF155, Cjun, Ini1, Cfos, CTCF, H3K36me3
		monocytes (Monocytes-CD14+)	CTCF, H3K36me3
		endothelium (HUVEC)	H3K36me3, H3K4me1, Max, Cjun, H3K27ac, PolII, CTCF, Cmyc, DNase1
		A549	H3K4me3, H3K4me1, H3K4me2, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00000400272 (promoter flanking region)	4	NHLF	DNase1, H3K4me1, H3K27ac, H3K36me3
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K4me2, DNase1
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K27ac, DNase1, H3K4me1
		muscle (HSMM)	H3K27ac, DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1, H3K36me3, H3K4me1, H3K27ac, H3K4me2, PolII
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun
		liver (HepG2)	H3K36me3, FOXA1, H3K4me1
		lung (IMR90)	H2BK120ac, H4K91ac, DNase1, H2BK12ac, H3K27ac, H3K36me3
		A549	H3K4me3, DNase1, H3K27ac, H3K36me3
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K27ac, H3K4me1
ENSR00001665285 (enhancer)	3	monocytes (Monocytes-CD14+)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00000400275 (promoter flanking region)	2	embryonic stem cell (H1ESC)	H3K36me3, Rad21, CTCF, DNase1
		HSMMtube	CTCF, DNase1
		blood (K562)	Rad21, CTCF, H3K4me1, H3K36me3, DNase1
		skin (NHDF-AD)	DNase1, CTCF

		muscle (HSMM)	H3K27ac, CTCF, H3K36me3, DNase1
		liver (HepG2)	FOSL2, Jund, HNF4G, HDAC2, Rad21, H3K4me1, p300, H3K4me2, HNF4A, FOXA1, H3K27ac, CTCF, Cmyc, H3K36me3, DNase1
		blood (GM12878)	CTCF
		nervous (NH-A)	DNase1, H3K36me3
		skin (NHEK)	H3K36me3, CTCF, H3K4me1, DNase1
		NHLF	H3K36me3, CTCF
		Osteobl	H3K36me3, CTCF
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF
		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, Cfos, H3K4me1, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, CTCF, DNase1, Cjun
		A549	H3K4me3, H3K4me2, CTCF, DNase1, H3K27ac, H3K36me3
ENSR00001448545	1	NHLF	DNase1
(promoter flanking region)		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	H3K27ac, DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K4me1, H3K36me3
		lung (IMR90)	DNase1
		nervous (NH-A)	DNase1
		A549	H3K36me3
ENSR00000400285	1	embryonic stem cell (H1ESC)	H3K36me3
(promoter flanking region)		HSMMtube	H3K36me3
		blood (K562)	H3K36me3
		skin (NHDF-AD)	DNase1, H3K4me1
		muscle (HSMM)	H3K36me3
		liver (HepG2)	H3K79me2, H3K4me1, H3K36me3
		lung (IMR90)	DNase1, H3K36me3, CTCF
		nervous (NH-A)	DNase1
		skin (NHEK)	H3K36me3
		NHLF	DNase1, H3K4me1
		Osteobl	H3K27ac, CTCF
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	DNase1, CTCF
		A549	H3K36me3

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
GDF5	downstream gene variant	3202	ENST00000374369	NM_000557.3	ENSP00000363489	3
GDF5	downstream gene variant	3202	ENST00000374372	?	ENSP00000363492	3
GDF5OS	upstream gene variant	2884	ENST00000374375	?	ENSP00000363495	3
UQCC1	upstream gene variant	4981	ENST00000498651	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374385	NM_018244.4	ENSP00000363506	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374380	NM_001184977.1	ENSP00000363501	4
UQCC1	upstream gene variant	3961	ENST00000491125	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374394	?	ENSP00000363515	4
UQCC1	downstream gene variant, upstream gene variant	385	ENST00000443429	?	ENSP00000416246	3
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000472559	?	?	5
UQCC1	upstream gene variant	3964	ENST00000491040	?	ENSP00000420584	1
UQCC1	downstream gene variant, upstream gene variant	886	ENST00000453855	?	ENSP00000390334	4

UQCC1	downstream gene variant, upstream gene variant	19	ENST00000457259	?	ENSP00000411024	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000349714	?	ENSP00000335364	4
UQCC1	downstream gene variant, upstream gene variant	162	ENST00000497717	?	?	3
UQCC1	downstream gene variant, upstream gene variant	907	ENST00000496812	?	?	5
UQCC1	downstream gene variant, upstream gene variant	455	ENST00000438533	?	ENSP00000398531	3
UQCC1	downstream gene variant, upstream gene variant	1424	ENST00000473982	?	?	4
UQCC1	upstream gene variant	528	ENST00000495752	?	?	1
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000359226	?	ENSP00000352161	4
UQCC1	upstream gene variant	3964	ENST00000397553	?	ENSP00000380685	1
UQCC1	downstream gene variant, upstream gene variant	1247	ENST00000482440	?	?	5
UQCC1	upstream gene variant	3963	ENST00000397554	?	ENSP00000380686	1
UQCC1	downstream gene variant, upstream gene variant	1444	ENST00000424405	?	ENSP00000399713	4
UQCC1	downstream gene variant, upstream gene variant	19	ENST00000374384	NM_199487.2	ENSP00000363505	4
UQCC1	downstream gene variant, upstream gene variant	840	ENST00000397556	?	ENSP00000380688	4

Putative effect on transcript

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5	ENST00000374369	NM_000557.3	ENSP00000363489	1
GDF5	ENST00000374372	?	ENSP00000363492	1
UQCC1	ENST00000397554	?	ENSP00000380686	7
UQCC1	ENST00000443429	?	ENSP00000416246	22
UQCC1	ENST00000472559	?	?	1
UQCC1	ENST00000374380	NM_001184977.1	ENSP00000363501	23
UQCC1	ENST00000374394	?	ENSP00000363515	23
UQCC1	ENST00000497717	?	?	15
UQCC1	ENST00000453855	?	ENSP00000390334	23
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UQCC1	ENST00000359226	?	ENSP00000352161	23
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UQCC1	ENST00000457259	?	ENSP00000411024	23
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UQCC1	ENST00000424405	?	ENSP00000399713	23
UQCC1	ENST00000374384	NM_199487.2	ENSP00000363505	23

UQCC1	ENST00000397556	?	ENSP00000380688	21

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5OS	ENST00000374375	?	ENSP00000363495	1