

Block annotations

Block info

genomic range	chr20:33,887,955-34,023,219 <i>e!</i>
block size	135,265 bp
variant count	39 variants

Basic features
























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

Trait annotations

Variant association

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

Disease gene annotation

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

## cis-eQTL

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

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

CPNE1	e!	ENST00000401607	e!							
CPNE1	e!	ENST00000498814	e!							
CPNE1	e!	ENST00000352393	e!							
CPNE1	e!	ENST00000483359	e!							
CPNE1	e!	ENST00000473373	e!							
CPNE1	e!	ENST00000483495	e!							
CPNE1	e!	ENST00000397443	e!	ILMN_2307025	e!	blood	1.05×10 <sup>-9</sup> (p-value)	MuTHER consortium		12
CPNE1	e!	ENST00000317677	e!			adipocyte	8.95×10 <sup>-8</sup> (p-value)	MuTHER consortium		12
CPNE1	e!	ENST00000498056	e!			skin	2.73×10 <sup>-5</sup> (p-value)	MuTHER consortium		12
CPNE1	e!	ENST00000437340	e!							
CPNE1	e!	ENST00000415920	e!							
CPNE1	e!	ENST00000397445	e!							
CPNE1	e!	ENST00000462352	e!							
CPNE1	e!	ENST00000401607	e!							
CPNE1	e!	ENST00000430570	e!							
CPNE1	e!	ENST00000352393	e!							
CPNE1	e!	ENST00000483359	e!							
CPNE1	e!	ENST00000483495	e!							
CPNE1	e!	ENST00000473373	e!							
GDF5	e!	?		ENSG00000125965	e!	muscularis mucosae	1.09×10 <sup>-5</sup> (p-value)	GTEX Portal V6		7
RPL36P4	e!	?		ENSG00000224497	e!	muscularis mucosae	2.23×10 <sup>-5</sup> (p-value)	GTEX Portal V6		7
UQCC1	e!	?		ENSG00000101019	e!	visceral adipocytes	1.02×10 <sup>-7</sup> (p-value)	GTEX Portal V6		12
GDF5	e!	?		ENSG00000125965	e!	sigmoid colon	6.58×10 <sup>-6</sup> (p-value)	GTEX Portal V6		9
UQCC1	e!	ENST00000374384	e!	ILMN_1745152	e!	monocyte	1.61×10 <sup>-24</sup> (p-value)	Zeller et al.		4
UQCC1	e!	ENST00000457259	e!							
UQCC1	e!	ENST00000359226	e!							
UQCC1	e!	ENST00000374380	e!							
UQCC1	e!	ENST00000472559	e!							
UQCC1	e!	ENST00000374394	e!							
UQCC1	e!	ENST00000349714	e!							
UQCC1	e!	ENST00000496812	e!							
UQCC1	e!	ENST00000374385	e!							
MMP24-AS1	e!	ENST00000566203	e!	ILMN_1911503	e!	blood	3.89×10 <sup>-6</sup> (p-value)	Westra et al.		11
CEP250	e!	ENST00000397527	e!	ILMN_1799011	e!	blood	1.12×10 <sup>-6</sup> (p-value)	Westra et al.		9
EDEM2	e!	ENST00000374491	e!	ILMN_1711909	e!	blood	7.10×10 <sup>-7</sup> (p-value)	Westra et al.		2
EDEM2	e!	ENST00000374492	e!							
UQCC1	e!	?		ENSG00000101019	e!	unexposed skin	1.72×10 <sup>-6</sup> (p-value)	GTEX Portal V6		3
PROCR	e!	?		ENSG00000101000	e!	pancreas	4.03×10 <sup>-6</sup> (p-value)	GTEX Portal V6		5
RPL36P4	e!	?		ENSG00000224497	e!	frontal cortex	2.41×10 <sup>-6</sup> (p-value)	GTEX Portal V6		19
RPL36P4	e!	?		ENSG00000224497	e!	transverse colon	1.77×10 <sup>-6</sup> (p-value)	GTEX Portal V6		10
UQCC1	e!	?		ENSG00000101019	e!	transverse colon	4.32×10 <sup>-6</sup> (p-value)	GTEX Portal V6		2
RPL36P4	e!	?		ENSG00000224497	e!	testis	1.43×10 <sup>-5</sup> (p-value)	GTEX Portal V6		4
UQCC1	e!	?		ENSG00000101019	e!	breast	8.32×10 <sup>-6</sup> (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 <i>e!</i>	1	ENCP00000031386	ERGIC3 <i>e!</i>
		ENCP00000031342	GSS <i>e!</i>
		ENCP00000031336	ACSS2 <i>e!</i>
		ENCP00000031392	FER1L4 <i>e!</i>
		ENCP00000031410	NFS1 <i>e!</i> ROMO1 <i>e!</i>
		ENCP00000031393	FER1L4 <i>e!</i>
		ENCP00000031390	FER1L4 <i>e!</i>
		ENCP00000031366	CEP250 <i>e!</i>
		ENCP00000031346	MYH7B <i>e!</i>
		ENCP00000031405	CPNE1 <i>e!</i> RBM12 <i>e!</i>
		ENCP00000031347	TRPC4AP <i>e!</i>

## Regulatory feature cluster

element id	variant(s)	tissue/cell	factors		
ENSR00001448537 <i>e!</i> (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 <i>e!</i> (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 <i>e!</i> (enhancer)	3	monocytes (Monocytes-CD14+)	H3K36me3		
		lung (IMR90)	H3K36me3		
		A549	H3K36me3		
ENSR00000400275 <i>e!</i> (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 <i>e!</i>	1 (promoter flanking region)	NHLF DNase1
	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 <i>e!</i>	1 (promoter flanking region)	embryonic stem cell (H1ESC) H3K36me3
	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 <i>e!</i>	downstream gene variant	3202	ENST00000374369 <i>e!</i>	NM_000557.3	ENSP00000363489 <i>e!</i>	3
GDF5 <i>e!</i>	downstream gene variant	3202	ENST00000374372 <i>e!</i>	?	ENSP00000363492 <i>e!</i>	3
GDF5OS <i>e!</i>	upstream gene variant	2884	ENST00000374375 <i>e!</i>	?	ENSP00000363495 <i>e!</i>	3
UQCC1 <i>e!</i>	upstream gene variant	4981	ENST00000498651 <i>e!</i>	?	?	1
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	19	ENST00000374385 <i>e!</i>	NM_018244.4	ENSP00000363506 <i>e!</i>	4
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	19	ENST00000374380 <i>e!</i>	NM_001184977.1	ENSP00000363501 <i>e!</i>	4
UQCC1 <i>e!</i>	upstream gene variant	3961	ENST00000491125 <i>e!</i>	?	?	1
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	19	ENST00000374394 <i>e!</i>	?	ENSP00000363515 <i>e!</i>	4
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	385	ENST00000443429 <i>e!</i>	?	ENSP00000416246 <i>e!</i>	3
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	19	ENST00000472559 <i>e!</i>	?	?	5
UQCC1 <i>e!</i>	upstream gene variant	3964	ENST00000491040 <i>e!</i>	?	ENSP00000420584 <i>e!</i>	1
UQCC1 <i>e!</i>	downstream gene variant, upstream gene variant	886	ENST00000453855 <i>e!</i>	?	ENSP00000390334 <i>e!</i>	4

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

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5 <i>e!</i>	ENST00000374369 <i>e!</i>	NM_000557.3	ENSP00000363489 <i>e!</i>	1
GDF5 <i>e!</i>	ENST00000374372 <i>e!</i>	?	ENSP00000363492 <i>e!</i>	1
UQCC1 <i>e!</i>	ENST00000397554 <i>e!</i>	?	ENSP00000380686 <i>e!</i>	7
UQCC1 <i>e!</i>	ENST00000443429 <i>e!</i>	?	ENSP00000416246 <i>e!</i>	22
UQCC1 <i>e!</i>	ENST00000472559 <i>e!</i>	?	?	1
UQCC1 <i>e!</i>	ENST00000374380 <i>e!</i>	NM_001184977.1	ENSP00000363501 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000374394 <i>e!</i>	?	ENSP00000363515 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000497717 <i>e!</i>	?	?	15
UQCC1 <i>e!</i>	ENST00000453855 <i>e!</i>	?	ENSP00000390334 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000397553 <i>e!</i>	?	ENSP00000380685 <i>e!</i>	7
UQCC1 <i>e!</i>	ENST00000349714 <i>e!</i>	?	ENSP00000335364 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000374385 <i>e!</i>	NM_018244.4	ENSP00000363506 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000496812 <i>e!</i>	?	?	1
UQCC1 <i>e!</i>	ENST00000438533 <i>e!</i>	?	ENSP00000398531 <i>e!</i>	22
UQCC1 <i>e!</i>	ENST00000473982 <i>e!</i>	?	?	3
UQCC1 <i>e!</i>	ENST00000491125 <i>e!</i>	?	?	7
UQCC1 <i>e!</i>	ENST00000359226 <i>e!</i>	?	ENSP00000352161 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000491040 <i>e!</i>	?	ENSP00000420584 <i>e!</i>	7
UQCC1 <i>e!</i>	ENST00000457259 <i>e!</i>	?	ENSP00000411024 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000495752 <i>e!</i>	?	?	3
UQCC1 <i>e!</i>	ENST00000424405 <i>e!</i>	?	ENSP00000399713 <i>e!</i>	23
UQCC1 <i>e!</i>	ENST00000374384 <i>e!</i>	NM_199487.2	ENSP00000363505 <i>e!</i>	23

UQCC1 *e!* ENST00000397556 *e!* ? ENSP00000380688 *e!* 21

**3'-UTR variant**

gene	affected transcript	RefSeq id	protein	variant(s)
GDF5OS <i>e!</i>	ENST00000374375 <i>e!</i>	?	ENSP00000363495 <i>e!</i>	1



