



HCG22	e!	?	ENSG00000228789	e!	coronary artery	1.07×10 <sup>-6</sup> (p-value)	GTE Portal V6	6	
LINC00243	e!	?	ENSG00000214894	e!	blood	2.13×10 <sup>-9</sup> (p-value)	GTE Portal V6	15	
C4A	e!	?	ENSG00000244731	e!	blood	2.49×10 <sup>-7</sup> (p-value)	GTE Portal V6	4	
MUC22	e!	?	ENSG00000261272	e!	esophagus mucosa	7.09×10 <sup>-9</sup> (p-value)	GTE Portal V6	15	
MICB	e!	?	ENSG00000204516	e!	esophagus mucosa	8.59×10 <sup>-7</sup> (p-value)	GTE Portal V6	5	
HLA-C	e!	?	ENSG00000204525	e!	skeletal muscle	7.95×10 <sup>-8</sup> (p-value)	GTE Portal V6	9	
C4A	e!	?	ENSG00000244731	e!	skeletal muscle	2.84×10 <sup>-7</sup> (p-value)	GTE Portal V6	4	
MICB	e!	?	ENSG00000204516	e!	cerebellum	6.88×10 <sup>-8</sup> (p-value)	GTE Portal V6	15	
CCHCR1	e!	?	ENSG00000204536	e!	transverse colon	4.24×10 <sup>-8</sup> (p-value)	GTE Portal V6	9	
CCHCR1	e!	?	ENSG00000204536	e!	subcutaneous adipocytes	1.68×10 <sup>-6</sup> (p-value)	GTE Portal V6	7	
HCG22	e!	?	ENSG00000228789	e!	subcutaneous adipocytes	2.19×10 <sup>-6</sup> (p-value)	GTE Portal V6	5	
HCG22	e!	ENST00000570223	e!	ILMN_1667229	e!	blood	3.23×10 <sup>-18</sup> (p-value)	MuTHER consortium	7
HCG22	e!	ENST00000565192	e!						
HCG22	e!	ENST00000615046	e!						
HCG22	e!	ENST00000426185	e!						
HCG22	e!	ENST00000562344	e!						
MICB	e!	ENST00000538442	e!	ILMN_1708006	e!	skin	1.20×10 <sup>-7</sup> (p-value)	MuTHER consortium	7
MICB	e!	ENST00000399150	e!						
MICB	e!	ENST00000252229	e!						
VARS2	e!	ENST00000321897	e!	ILMN_1737585	e!	skin	2.72×10 <sup>-5</sup> (p-value)	MuTHER consortium	1
VARS2	e!	ENST00000625423	e!						
VARS2	e!	ENST00000541562	e!						
VARS2	e!	ENST00000542001	e!						
VARS2	e!	ENST00000469358	e!						
VARS2	e!	ENST00000477288	e!						
VARS2	e!	ENST00000473916	e!						
VARS2	e!	ENST00000476162	e!						
HCP5	e!	ENST00000414046	e!	ILMN_1803945	e!	skin	3.52×10 <sup>-5</sup> (p-value)	MuTHER consortium	2
HCP5	e!	ENST00000541196	e!						
FLOT1	e!	?	ENSG00000137312	e!	thyroid	8.04×10 <sup>-6</sup> (p-value)	GTE Portal V6	1	
C4A	e!	?	ENSG00000244731	e!	thyroid	4.85×10 <sup>-6</sup> (p-value)	GTE Portal V6	1	
?	?	?	ILMN_1679389	e!	b-cell	3.75×10 <sup>-4</sup> (p-value)	Fairfax et al.	3	
C4A	e!	?	ENSG00000244731	e!	atrial appendage	2.58×10 <sup>-7</sup> (p-value)	GTE Portal V6	5	
C4A	e!	?	ENSG00000244731	e!	tibial nerve	3.53×10 <sup>-7</sup> (p-value)	GTE Portal V6	9	
C4A	e!	?	ENSG00000244731	e!	putamen	2.07×10 <sup>-7</sup> (p-value)	GTE Portal V6	7	
C4A	e!	?	ENSG00000244731	e!	left ventricle	3.19×10 <sup>-7</sup> (p-value)	GTE Portal V6	9	
C4A	e!	?	ENSG00000244731	e!	muscularis mucosae	1.19×10 <sup>-8</sup> (p-value)	GTE Portal V6	8	
CYP21A1P	e!	?	ENSG00000204338	e!	muscularis mucosae	2.21×10 <sup>-7</sup> (p-value)	GTE Portal V6	2	
HLA-C	e!	ENST00000470363	e!	216526_x_at	e!	blood	6.20×10 <sup>-8</sup> (p-value)	Dixon et al.	2
HLA-C	e!	ENST00000383329	e!						
HLA-C	e!	ENST00000487245	e!						
HLA-C	e!	ENST00000376228	e!						

HLA-C <i>e!</i>	ENST00000466892 <i>e!</i>						
HLA-C <i>e!</i>	ENST00000376237 <i>e!</i>						
HLA-C <i>e!</i>	ENST00000620806 <i>e!</i>						
?	?	ILMN_1893395 <i>e!</i>	monocyte	8.28×10 <sup>-18</sup> (p-value)	Zeller et al. <i>!M</i>	2	
?	?	ILMN_1883997 <i>e!</i>	monocyte	7.04×10 <sup>-18</sup> (p-value)	Zeller et al. <i>!M</i>	2	
C4A <i>e!</i>	?	ENSG00000244731 <i>e!</i>	transformed fibroblasts	3.34×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1	
C4A <i>e!</i>	?	ENSG00000244731 <i>e!</i>	sun exposed skin	1.74×10 <sup>-7</sup> (p-value)	GTEx Portal V6 <i>!M</i>	2	
C4A <i>e!</i>	?	ENSG00000244731 <i>e!</i>	unexposed skin	1.13×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1	
HLA-C <i>e!</i>	?	ENSG00000204525 <i>e!</i>	EBV lymphocytes	3.03×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1	
CYP21A1P <i>e!</i>	?	ENSG00000204338 <i>e!</i>	aorta	2.61×10 <sup>-6</sup> (p-value)	GTEx Portal V6 <i>!M</i>	1	

**trans-eQTL**

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	221491_x_at <i>e!</i>	chr6	blood	6.20×10 <sup>-8</sup> (p-value)	Dixon et al. <i>!M</i>	2

**Putative effect on regulation**

**ENCODE promoter-associated distal DHS (Enhancer)**

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000444432 <i>e!</i>	1	ENCP00000047915	PPP1R10 <i>e!</i> PPP1R10 <i>e!</i> PPP1R10 <i>e!</i> PPP1R10 <i>e!</i> PPP1R10 <i>e!</i> PPP1R10 <i>e!</i>
		ENCP00000047967	VARS2 <i>e!</i> VARS2 <i>e!</i> VARS2 <i>e!</i> VARS2 <i>e!</i> VARS2 <i>e!</i> VARS2 <i>e!</i>
		ENCP00000047911	PRR3 <i>e!</i> PRR3 <i>e!</i> PRR3 <i>e!</i> GNL1 <i>e!</i> GNL1 <i>e!</i> GNL1 <i>e!</i> PRR3 <i>e!</i> GNL1 <i>e!</i> PRR3 <i>e!</i> GNL1 <i>e!</i> GNL1 <i>e!</i> GNL1 <i>e!</i> PRR3 <i>e!</i> PRR3 <i>e!</i> GNL1 <i>e!</i>
		ENCP00000047978	CDSN <i>e!</i> CDSN <i>e!</i> CDSN <i>e!</i> CDSN <i>e!</i> CDSN <i>e!</i> CDSN <i>e!</i>
		ENCP00000047992	HCG27 <i>e!</i> HCG27 <i>e!</i> HCG27 <i>e!</i>

			HCG27 <i>e!</i> HCG27 <i>e!</i> HCG27 <i>e!</i> HCG27 <i>e!</i>
		ENCP00000047974	XXbac-BPG118E17.9 <i>e!</i> XXbac-BPG118E17.9 <i>e!</i> XXbac-BPG118E17.9 <i>e!</i> XXbac-BPG118E17.9 <i>e!</i> XXbac-BPG118E17.9 <i>e!</i> XXbac-BPG118E17.9 <i>e!</i>
		ENCP00000047923	ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i>
		ENCP00000047975	HCG22 <i>e!</i> HCG22 <i>e!</i> HCG22 <i>e!</i> HCG22 <i>e!</i> HCG22 <i>e!</i> HCG22 <i>e!</i> HCG22 <i>e!</i>
		ENCP00000047995	XXbac-BPG299F13.16 <i>e!</i>
		ENCP00000048019	MICB <i>e!</i> MICB <i>e!</i> MICB <i>e!</i> MICB <i>e!</i> MICB <i>e!</i> MICB <i>e!</i> MICB <i>e!</i>
ENSE00000444471 <i>e!</i>	1	ENCP00000048001	XXbac-BPG248L24.13 <i>e!</i>
		ENCP00000047921	ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i> ATAT1 <i>e!</i>
		ENCP00000047949	DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i> DDR1 <i>e!</i>
		ENCP00000047941	FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i> FLOT1 <i>e!</i>

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001703009 <i>e!</i> (open chromatin region)	2	blood (K562)	H3K27me3
ENSR00001703011 <i>e!</i> (CTCF binding site)	1	NHLF embryonic stem cell (H1ESC) HSMMtube Osteobl blood (K562) blood (DND-41)	DNase1 H3K27me3, Rad21, CTCF, DNase1 DNase1, H3K27me3 H3K4me2 H3K27me3 H3K27me3

ENSR00001494536 <i>e!</i> (promoter flanking region)	1	blood (DND-41) skin (NHDF-AD) breast (HMEC) muscle (HSMM) cervix (HeLa-S3) monocytes (Monocytes-CD14+) endothelium (HUVEC) liver (HepG2) lung (IMR90) nervous (NH-A)	H3K27me3 CTCF, H3K4me3, DNase1, H3K4me2 CTCF, H3K4me2 DNase1 CTCF H3K27me3 H3K27me3 CTCF, H3K27me3 H3K4me2 H3K4me2, DNase1
		embryonic stem cell (H1ESC) HSMMtube blood (K562) skin (NHDF-AD) muscle (HSMM) liver (HepG2) blood (GM12878) lung (IMR90) nervous (NH-A) NHLF Osteobl blood (DND-41) breast (HMEC) cervix (HeLa-S3) monocytes (Monocytes-CD14+) endothelium (HUVEC)	DNase1, CTCF, H3K27me3, Rad21 DNase1, H3K27me3 H3K27me3 CTCF, H3K4me3, DNase1, H3K4me2 DNase1 CTCF, H3K27me3 H3K27me3 H3K4me2, H3K27me3 H3K4me2, DNase1 DNase1 H3K4me2 H3K27me3 H3K4me2, CTCF CTCF H3K27me3 H3K27me3

#### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
MUC21 <i>e!</i>	upstream gene variant	475	ENST00000376296 <i>e!</i>	NM_001010909.2	ENSP00000365473 <i>e!</i>	2
MUC21 <i>e!</i>	upstream gene variant, downstream gene variant	475	ENST00000486149 <i>e!</i>	?	ENSP00000457640 <i>e!</i>	3

#### Putative effect on transcript

#### Synonymous coding variant

gene	affected transcript	RefSeq id	protein	AA's	exchanged codons	variant(s)
MUC21 <i>e!</i>	ENST00000376296 <i>e!</i>	NM_001010909.2	ENSP00000365473 <i>e!</i>	T	acA/acG	1

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC22 <i>e!</i>	ENST00000561890 <i>e!</i>	NM_001198815.1	ENSP00000455906 <i>e!</i>	2

#### 3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21 <i>e!</i>	ENST00000376296 <i>e!</i>	NM_001010909.2	ENSP00000365473 <i>e!</i>	1

#### 5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
MUC21 <i>e!</i>	ENST00000486149 <i>e!</i>	?	ENSP00000457640 <i>e!</i>	1

