SNiPAcard

Block annotations

Block info

genomic range	chr20:34,199,977-34,562,924
block size	362,948 bp
variant count	168 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.159 [-3.59 – 9.577]	**gene(s) hit or close-by**	CNBD2, COX7BP2, CPNE1, FER1L4, HIGD1AP16, NFS1, PHF20, RBM12, RBM39, RN7SKP271, RNU4-40P, RNU6-759P, RNU6-937P, ROMO1, RP1-309K20.6, RPF2P1, SCAND1, SPAG4, Y_RNA
phastCons	μ = 0.080 [0 – 1]	**eQTL gene(s)**	CEP250, CPNE1, EPB41L1, ERGIC3, FER1L4, LINC00657, MMP24-AS1, RBM39, RPL36P4, SCAND1
GERP++	μ = -0.096 [-5.54 – 5.32]	**potentially regulated gene(s)**	CEP250, CPNE1, EPB41L1, ERGIC3, FER1L4, GDF5, HMGB3P2, NFS1, PHF20, RBM12, RBM39, ROMO1, RP11-234K24.3, RP3-477O4.14, RP3-477O4.5, RP4-550H1.4, RP4-550H1.5, SCAND1, SPAG4
CADD score	μ = 4.564 [0.041 – 22.4]	**disease gene(s)**	EPB41L1, GDF5, NFS1

Trait annotations

Disease gene annotation

gene	trait	source DB	source entry/link
EPB41L1	MENTAL RETARDATION, AUTOSOMAL DOMINANT 11	OMIM	MIM:614257
EPB41L1	Autosomal dominant non-syndromic intellectual disability	OrphaNet	OrphaNet:178469
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
NFS1	Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency	OrphaNet	OrphaNet:397593
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

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)
CPNE1	missense variant	ENST00000352393	?	ENSP00000336945	R/P	cGt/cCt	?	?	1

CPNE1	missense variant	ENST00000317677	NM_003915.5	ENSP00000317257	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000401607	?	ENSP00000386067	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397442	?	ENSP00000380584	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000430570	?	ENSP00000390626	R/P	cGt/cCt	?	?	1
CPNE1	missense variant	ENST00000437340	?	ENSP00000415597	R/P	cGt/cCt	?	?	1
PHF20	missense variant	ENST00000439301	?	ENSP00000410373	M/V	Atg/Gtg	?	?	1
PHF20	missense variant	ENST00000374012	NM_016436.4	ENSP00000363124	M/V	Atg/Gtg	?	?	1

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
FER1L4	?	ENSG00000088340	atrial appendage	3.70×10^{-8} (p-value)	GTEx Portal V6	156
RPL36P4	?	ENSG00000224497	atrial appendage	1.15×10^{-10} (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	atrial appendage	1.91×10^{-46} (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	transformed fibroblasts	1.75×10^{-8} (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	transformed fibroblasts	8.96×10^{-18} (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	transformed fibroblasts	4.53×10^{-66} (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	breast	3.38×10^{-8} (p-value)	GTEx Portal V6	154
CPNE1	?	ENSG00000214078	breast	2.47×10^{-36} (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	blood	2.32×10^{-11} (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	blood	2.41×10^{-71} (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	blood	4.47×10^{-7} (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	skeletal muscle	3.85×10^{-14} (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	skeletal muscle	1.36×10^{-7} (p-value)	GTEx Portal V6	125
CPNE1	?	ENSG00000214078	skeletal muscle	2.47×10^{-46} (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	skeletal muscle	7.89×10^{-6} (p-value)	GTEx Portal V6	24
CPNE1	?	ENSG00000214078	putamen	9.62×10^{-14} (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	EBV lymphocytes	1.45×10^{-23} (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	EBV lymphocytes	6.76×10^{-7} (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	unexposed skin	6.74×10^{-13} (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	unexposed skin	4.43×10^{-14} (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	unexposed skin	7.48×10^{-40} (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	sun exposed skin	1.04×10^{-11} (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	sun exposed skin	9.86×10^{-16} (p-value)	GTEx Portal V6	167

CPNE1	?	ENSG00000214078	sun exposed skin	7.80×10-61 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	ovary	5.31×10-10 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	ovary	2.42×10-6 (p-value)	GTEx Portal V6	23
FER1L4	?	ENSG00000088340	left ventricle	9.16×10-11 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	left ventricle	2.47×10-8 (p-value)	GTEx Portal V6	165
CPNE1	?	ENSG00000214078	left ventricle	8.69×10-37 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	aorta	1.46×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	aorta	6.55×10-47 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	aorta	7.36×10-6 (p-value)	GTEx Portal V6	12
FER1L4	?	ENSG00000088340	subcutaneous adipocytes	1.21×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	subcutaneous adipocytes	5.85×10-16 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	subcutaneous adipocytes	2.74×10-64 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	caudate basal ganglia	4.34×10-15 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	visceral adipocytes	1.31×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	visceral adipocytes	5.53×10-49 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	stomach	8.62×10-10 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	stomach	1.47×10-35 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cortex	1.60×10-13 (p-value)	GTEx Portal V6	166
MMP24-AS1	?	ENSG00000126005	cortex	7.02×10-6 (p-value)	GTEx Portal V6	1
FER1L4	?	ENSG00000088340	tibial nerve	1.91×10-17 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	tibial nerve	2.79×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	tibial nerve	3.76×10-69 (p-value)	GTEx Portal V6	167
CEP250	?	ENSG00000126001	tibial nerve	4.38×10-6 (p-value)	GTEx Portal V6	56
RPL36P4	?	ENSG00000224497	pituitary	8.84×10-6 (p-value)	GTEx Portal V6	72
CPNE1	?	ENSG00000214078	pituitary	2.52×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	anterior cingulate cortex	5.59×10-15 (p-value)	GTEx Portal V6	166
ERGIC3	?	ENSG00000125991	anterior cingulate cortex	1.14×10-5 (p-value)	GTEx Portal V6	9
EPB41L1	?	ENSG00000088367	esophagus mucosa	1.24×10-7 (p-value)	GTEx Portal V6	161
RPL36P4	?	ENSG00000224497	esophagus mucosa	4.55×10-12 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	esophagus mucosa	1.19×10-71 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	testis	2.75×10-8 (p-value)	GTEx Portal V6	111
RPL36P4	?	ENSG00000224497	testis	1.05×10-8 (p-value)	GTEx Portal V6	165
CPNE1	?	ENSG00000214078	testis	1.01×10-36 (p-value)	GTEx Portal V6	167
LINC00657	?	ENSG00000260032	testis	1.47×10-5 (p-value)	GTEx Portal V6	9
CPNE1	?	ENSG00000214078	hypothalamus	1.38×10-11 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	frontal cortex	6.74×10-14 (p-value)	GTEx Portal V6	166

CPNE1	?	ENSG00000214078	uterus	7.38×10-9 (p-value)	GTEx Portal V6	162
RPL36P4	?	ENSG00000224497	uterus	2.48×10-7 (p-value)	GTEx Portal V6	5
RPL36P4	?	ENSG00000224497	pancreas	9.94×10-12 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	pancreas	2.51×10-38 (p-value)	GTEx Portal V6	167
RBM39	?	ENSG00000131051	pancreas	6.07×10-6 (p-value)	GTEx Portal V6	5
FER1L4	?	ENSG00000088340	muscularis mucosae	1.73×10-13 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	muscularis mucosae	1.28×10-14 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	muscularis mucosae	5.11×10-43 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	lung	4.47×10-20 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	lung	2.11×10-71 (p-value)	GTEx Portal V6	167
FER1L4	?	ENSG00000088340	tibial artery	2.42×10-11 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	tibial artery	3.68×10-13 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	tibial artery	6.92×10-60 (p-value)	GTEx Portal V6	167
EPB41L1	?	ENSG00000088367	tibial artery	1.87×10-6 (p-value)	GTEx Portal V6	32
FER1L4	?	ENSG00000088340	thyroid	4.16×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	thyroid	2.29×10-16 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	thyroid	1.79×10-67 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	prostate	9.68×10-13 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	prostate	6.99×10-7 (p-value)	GTEx Portal V6	1
RPL36P4	?	ENSG00000224497	transverse colon	2.04×10-11 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	transverse colon	4.93×10-49 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	terminal ileum	1.26×10-6 (p-value)	GTEx Portal V6	52
CPNE1	?	ENSG00000214078	terminal ileum	4.62×10-19 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	nucleus accumbens	1.11×10-7 (p-value)	GTEx Portal V6	165
RPL36P4	?	ENSG00000224497	nucleus accumbens	8.13×10-6 (p-value)	GTEx Portal V6	70
RPL36P4	?	ENSG00000224497	spleen	1.70×10-6 (p-value)	GTEx Portal V6	113
CPNE1	?	ENSG00000214078	spleen	2.05×10-21 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	coronary artery	4.85×10-10 (p-value)	GTEx Portal V6	155
CPNE1	?	ENSG00000214078	coronary artery	7.97×10-23 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	sigmoid colon	7.42×10-7 (p-value)	GTEx Portal V6	101
CPNE1	?	ENSG00000214078	sigmoid colon	5.99×10-20 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	vagina	3.60×10-12 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	gastroesophageal junction	7.44×10-10 (p-value)	GTEx Portal V6	166
CPNE1	?	ENSG00000214078	gastroesophageal junction	6.13×10-31 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cerebellar hemisphere	5.12×10-8 (p-value)	GTEx Portal V6	166
RPL36P4	?	ENSG00000224497	cerebellar hemisphere	8.05×10-6 (p-value)	GTEx Portal V6	1

FER1L4	?	ENSG00000088340	adrenal gland	1.94×10-8 (p-value)	GTEx Portal V6	167
RPL36P4	?	ENSG00000224497	adrenal gland	2.04×10-10 (p-value)	GTEx Portal V6	158
CPNE1	?	ENSG00000214078	adrenal gland	5.78×10-34 (p-value)	GTEx Portal V6	167
CPNE1	?	ENSG00000214078	cerebellum	2.33×10-9 (p-value)	GTEx Portal V6	166
CEP250	?	ENSG00000126001	cerebellum	1.89×10-5 (p-value)	GTEx Portal V6	21
CPNE1	?	ENSG00000214078	hippocampus	7.13×10-9 (p-value)	GTEx Portal V6	164
CPNE1	?	ENSG00000214078	liver	4.29×10-16 (p-value)	GTEx Portal V6	149
SCAND1	ENST00000373991	ILMN_1795317	monocyte	4.35×10-7 (p-value)	Fairfax et al.	13
SCAND1	ENST00000305978	ILMN_2372011	monocyte	2.41×10-6 (p-value)	Fairfax et al.	11
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	1.48×10-51 (p-value)	Fairfax et al.	13
CPNE1	ENST00000317677		b-cell	1.45×10-18 (p-value)	Fairfax et al.	13
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_2307025	monocyte	5.23×10-40 (p-value)	Fairfax et al.	13
CPNE1	ENST00000317677		b-cell	2.28×10-23 (p-value)	Fairfax et al.	13
CPNE1	ENST00000498056					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000437340					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	ILMN_1670841	skin	1.64×10-29 (p-value)	MuTHER consortium	52
CPNE1	ENST00000317677		blood	8.08×10-69 (p-value)	MuTHER consortium	52
CPNE1	ENST00000498056		adipocyte	4.98×10-34 (p-value)	MuTHER consortium	52
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	skin	1.95×10^{-30} (p-value)	MuTHER consortium	52
CPNE1	ENST00000317677		blood	9.52×10^{-70} (p-value)	MuTHER consortium	52
CPNE1	ENST00000498056		adipocyte	8.91×10^{-43} (p-value)	MuTHER consortium	52
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
SCAND1	ENST00000305978	ILMN_1794230	blood	1.14×10^{-5} (p-value)	MuTHER consortium	36
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
FER1L4	ENST00000615531	ILMN_2206722	blood	4.69×10^{-5} (p-value)	MuTHER consortium	1
FER1L4	ENST00000621044					
FER1L4	ENST00000616283					
FER1L4	ENST00000616711					
FER1L4	ENST00000611673					
FER1L4	ENST00000613061					
SCAND1	ENST00000305978	ILMN_2372011	blood	1.54×10^{-5} (p-value)	MuTHER consortium	32
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CEP250	ENST00000397527	ILMN_1799011	blood	1.64×10^{-5} (p-value)	Westra et al.	17
CPNE1	?	ENSG00000214078	blood	3.42×10^{-21} (q-value)	SeeQTL DB (HapMap)	23
SCAND1	ENST00000373991	ILMN_1795317	monocyte	7.72×10^{-41} (p-value)	Zeller et al.	10
SCAND1	ENST00000305978	ILMN_1794230	monocyte	1.59×10^{-13} (p-value)	Zeller et al.	10
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
SCAND1	ENST00000305978	ILMN_2372011	monocyte	8.37×10^{-17} (p-value)	Zeller et al.	10
SCAND1	ENST00000373991					
SCAND1	ENST00000615116					
CPNE1	ENST00000397443	ILMN_1670841	monocyte	1.80×10^{-150} (p-value)	Zeller et al.	10
CPNE1	ENST00000317677					
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	monocyte	1.72×10-116 (p-value)	Zeller et al.	10
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000397445					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000483495					
CPNE1	ENST00000473373					
CPNE1	ENST00000397443	ILMN_1670841	liver	<2.2×10-16 (p-value)	Innocenti et al.	1
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397442					
CPNE1	ENST00000437340					
CPNE1	ENST00000397445					
CPNE1	ENST00000415920					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000498814					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					
CPNE1	ENST00000397443	206918_s_at	blood	1.30×10-33 (p-value)	Dixon et al.	1
CPNE1	ENST00000317677					
CPNE1	ENST00000498056					
CPNE1	ENST00000397445					
CPNE1	ENST00000437340					
CPNE1	ENST00000415920					
CPNE1	ENST00000462352					
CPNE1	ENST00000401607					
CPNE1	ENST00000430570					
CPNE1	ENST00000352393					
CPNE1	ENST00000483359					
CPNE1	ENST00000473373					
CPNE1	ENST00000483495					

Putative effect on regulation

FANTOM5 expressed promoter

SNiPA promoter id	variant(s)	associated transcript(s)	gene
FFCP00000447127	1	ENST00000397443, ENST00000430570, ENST00000439806, ENST00000397445	CPNE1
		ENST00000435161	RBM12

ENCODE promoter-associated DHS

SNiPA promoter id	variant(s)	associated gene(s)
ENCP00000031405		RBM12
ENCP00000031420		

ENCP00000031420

ENCP00000031435

ENCODE promoter-associated distal DHS (Enhancer)

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000293219	1	ENCP00000031365	GDF5
		ENCP00000031373	RP3-477O4.14
		ENCP00000031370	CEP250
		ENCP00000031438	RP4-550H1.4
ENCE00000293515	1	ENCP00000031431	PHF20
ENCE00000293353	1	ENCP00000031397	SPAG4
		ENCP00000031415	RBM39
		ENCP00000031440	RP4-550H1.5
ENCE00000293517	1	ENCP00000031431	PHF20
ENCE00000293226	1	ENCP00000031446	RP11-234K24.3
		ENCP00000031448	EPB41L1
		ENCP00000031444	EPB41L1
		ENCP00000031410	NFS1 ROMO1
		ENCP00000031385	ERGIC3
		ENCP00000031367	CEP250
		ENCP00000031393	FER1L4
		ENCP00000031380	ERGIC3
		ENCP00000031378	RP3-477O4.5
		ENCP00000031406	NFS1
		ENCP00000031387	FER1L4
		ENCP00000031398	SPAG4
		ENCP00000031389	FER1L4
		ENCP00000031439	HMGB3P2
		ENCP00000031445	EPB41L1
		ENCP00000031392	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031443	EPB41L1
		ENCP00000031366	CEP250
		ENCP00000031381	ERGIC3
		ENCP00000031450	EPB41L1
		ENCP00000031427	PHF20
		ENCP00000031405	CPNE1 RBM12
ENCE00000293228	1	ENCP00000031439	HMGB3P2
		ENCP00000031446	RP11-234K24.3
		ENCP00000031392	FER1L4
		ENCP00000031393	FER1L4
		ENCP00000031390	FER1L4
		ENCP00000031366	CEP250
		ENCP00000031378	RP3-477O4.5
		ENCP00000031406	NFS1
		ENCP00000031387	FER1L4
		ENCP00000031450	EPB41L1
ENCE00000293397	1	ENCP00000031411	RBM39
ENCE00000293217	1	ENCP00000031364	GDF5
ENCE00000293297	1	ENCP00000031376	CEP250
ENCE00000293340	1	ENCP00000031394	FER1L4

	ENCP00000031395	FER1L4
	ENCP00000031442	EPB41L1

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001665301 (promoter)	1	embryonic stem cell (H1ESC)	H3K27me3, PolII, Rad21, TAF7, H3K9ac, CTCF, TAF1, H3K4me2, SP1, USF1, H3K4me3, DNase1
		HSMMtube	H3K4me2, H2AZ, DNase1
		blood (K562)	H2AZ, PolII, H3K4me2, CTCF, DNase1, H3K4me3, HEY1, H3K9ac, E2F6, Max, ZBTB7A, Rad21
		skin (NHDF-AD)	H3K4me2, DNase1, H3K4me3, CTCF
		muscle (HSMM)	H3K27ac, CTCF, H2AZ, DNase1
		liver (HepG2)	H3K36me3, H3K27me3, H3K4me3, Sin3Ak20, PolII, H3K79me2, TAF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, DNase1
		blood (GM12878)	H3K9ac, CTCF, H3K4me2, EBF1, PolII, BATF, H2AZ, DNase1, H3K4me3, H3K27ac
		lung (IMR90)	DNase1, H3K4me2, H3K4me3, CTCF
		nervous (NH-A)	DNase1, H3K9ac, H3K4me2
		skin (NHEK)	DNase1, H3K36me3, CTCF, H3K4me3, H3K4me2
		NHLF	DNase1, H3K9ac, H3K27ac
		Osteobl	H3K27ac, H3K4me2
		blood (DND-41)	H3K27me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, PolII, H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, Ini1, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K27me3, H3K4me3
		endothelium (HUVEC)	CTCF, DNase1, PolII, H3K4me1, H3K4me2, Max, H3K4me3
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00000400325 (promoter)	2	embryonic stem cell (H1ESC)	H3K9ac, H3K4me3, H4K5ac, H3K36me3, CTCF, PolII, H4K20me1, H3K27ac, Yy1, TAF1, H3K4me2, DNase1
		HSMMtube	H2AZ, DNase1, H3K4me3, H3K79me2, H3K9ac, H4K20me1, H3K4me2, H3K27ac
		blood (K562)	Brg1, Egr1, H3K27ac, Max, H3K9ac, Cmyc, H3K79me2, HEY1, Yy1, H2AZ, TAF1, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H3K79me2
		liver (HepG2)	H3K27ac, Cmyc, H3K4me3, H3K27me3, DNase1, H3K9ac, H3K4me2, Yy1, H4K20me1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ
		blood (GM12878)	H3K79me2, TAF1, PolII, H2AZ, DNase1, Yy1, ELF1, H3K4me3, H3K4me2, H3K9ac, H3K36me3, H3K27ac
		lung (IMR90)	H3K9ac, H3K4ac, DNase1, H4K8ac, H4K20me1, H3K79me2, H3K27ac, H3K4me2, H4K5ac, H3K4me3
		nervous (NH-A)	DNase1, H3K27ac, H3K4me3, H3K4me2, H3K9ac, H4K20me1
		skin (NHEK)	DNase1, H3K4me3, H3K4me2, H3K9ac, H3K27ac
		NHLF	DNase1, H3K4me3, H3K9ac, H3K27ac
		Osteobl	H4K20me1, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K27ac, DNase1, H3K4me3, H3K9ac, H3K4me2
		cervix (HeLa-S3)	DNase1, Nrf1, Ini1, H3K9ac, H3K4me2, H3K27ac, H3K36me3, PolII, H3K79me2, H3K4me3, TAF1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		endothelium (HUVEC)	H3K36me3, DNase1, PolII, Max, H3K4me2, H3K4me3, H3K9ac
		A549	H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K9ac, DNase1
ENSR00000400329 (promoter)	1	embryonic stem cell (H1ESC)	H3K4me3, H3K9ac, H3K36me3, CTCF, PolII, Rad21, Gabp, Sin3Ak20, H3K27ac, TAF1, H3K4me2, DNase1
		HSMMtube	H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H2AZ, ELF1, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3, H3K27ac, Max, CTCF, H3K9ac, Gabp, H3K79me2
		skin (NHDF-AD)	H3K4me3, DNase1, H3K27ac, H3K4me2, H3K9ac
		muscle (HSMM)	H3K36me3, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ
		liver (HepG2)	H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K27me3, H3K36me3, DNase1, PolII, Gabp
		blood (GM12878)	Tr4, H3K36me3, H3K9ac, H3K4me2, PolII, H2AZ, DNase1, CTCF, Gabp, ELF1, H3K4me3, H3K27ac
		lung (IMR90)	H3K27ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K4me3, H3K27ac, H3K36me3, DNase1
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H3K27ac, CTCF
		NHLF	H3K4me3, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K4me2, H3K4me3, H3K27ac
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, H3K4me2, H3K4me1
		breast (HMEC)	H3K27ac, CTCF, H3K36me3, H3K4me2, H3K9ac, H3K4me3

		cervix (HeLa-S3)	DNase1, H3K36me3, CTCF, PolII, Gabp, H3K4me3, TAF1, H3K27ac, H3K4me2, H3K9ac
		monocytes (Monocytes-CD14+)	H3K4me3, H3K36me3, H3K9ac, DNase1, H3K4me1, H3K4me2, H3K27ac
		endothelium (HUVEC)	Max, H3K4me3, H3K4me2, H3K9ac, PolII, DNase1, H3K36me3
		A549	H3K36me3, H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac
ENSR00000400333 (open chromatin region)	1	embryonic stem cell (H1ESC)	H3K36me3
		blood (K562)	H3K36me3
		muscle (HSMM)	H3K36me3
		breast (HMEC)	H3K36me3
		endothelium (HUVEC)	H3K36me3
		blood (GM12878)	PolII, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001404744 (open chromatin region)	2	embryonic stem cell (H1ESC)	PolII, H3K36me3
		blood (K562)	H3K36me3, H3K79me2
		breast (HMEC)	H3K36me3
		muscle (HSMM)	H3K79me2, H3K36me3
		endothelium (HUVEC)	H3K36me3
		lung (IMR90)	H4K20me1, H3K36me3
		blood (GM12878)	H3K36me3, H3K79me2
		A549	H3K36me3
		skin (NHEK)	H3K36me3, DNase1
ENSR00001665311 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, PolII
		HSMMtube	H3K36me3
		blood (K562)	H3K79me2
		muscle (HSMM)	H3K36me3, H3K79me2
		liver (HepG2)	H4K20me1, PolII, H3K79me2, H3K4me1, H3K36me3
		blood (GM12878)	PolII, H3K79me2, H3K4me3, H3K36me3
		lung (IMR90)	H3K36me3
		nervous (NH-A)	H3K36me3
		skin (NHEK)	H3K36me3
		NHLF	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	H3K36me3, PolII
		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		A549	H3K36me3
ENSR00000400351 (enhancer)	1	lung (IMR90)	H3K27me3
ENSR00001448569 (promoter flanking region)	1	embryonic stem cell (H1ESC)	CTCF
		HSMMtube	DNase1
		liver (HepG2)	H3K27me3
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	DNase1
		muscle (HSMM)	DNase1, H3K27ac
ENSR00001665314 (enhancer)	1	liver (HepG2)	H3K27me3
		lung (IMR90)	H3K27me3
ENSR00000234313 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		blood (K562)	DNase1, H3K36me3
		muscle (HSMM)	H3K36me3
		cervix (HeLa-S3)	H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K79me2, H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
ENSR00001665315 (enhancer)	2	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		blood (DND-41)	H3K36me3
		blood (K562)	Egr1
		breast (HMEC)	H3K36me3
		cervix (HeLa-S3)	H3K36me3

		monocytes (Monocytes-CD14+)	H4K20me1, H3K36me3
		endothelium (HUVEC)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00001448575 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	DNase1
		Osteobl	H3K36me3, H3K27ac
		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K27ac, DNase1
		muscle (HSMM)	H3K27ac, H3K36me3, DNase1
		cervix (HeLa-S3)	H3K36me3, DNase1, Jund, Cjun
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		A549	H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000400367 (promoter flanking region)	1	NHLF	DNase1
		embryonic stem cell (H1ESC)	H3K36me3
		HSMMtube	DNase1
		Osteobl	H3K36me3
		blood (DND-41)	H3K36me3
		muscle (HSMM)	DNase1, H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		liver (HepG2)	FOXA1, H3K36me3
		blood (GM12878)	H3K36me3
		nervous (NH-A)	DNase1
		A549	DNase1, H3K36me3
		skin (NHEK)	H3K36me3
ENSR00000400368 (TF binding site)	1	lung (IMR90)	H3K36me3
ENSR00000400373 (promoter flanking region)	3	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K27ac, H3K36me3, DNase1
		blood (K562)	DNase1, H3K36me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3, H3K27ac
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	H3K36me3, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	H3K36me3, H3K4me2, H3K9ac, H3K27ac, H3K4me1, DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K36me3, H3K4me2, H3K27ac
		blood (DND-41)	H3K36me3
		breast (HMEC)	H3K27ac, DNase1
		cervix (HeLa-S3)	H3K27ac, H3K4me2, Jund, PolII, TAF1, Max, Cfos, DNase1, H3K36me3
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	DNase1, Cjun
		A549	H3K36me3, DNase1, H3K4me2, H3K4me3
ENSR00000400377 (promoter flanking region)	1	embryonic stem cell (H1ESC)	DNase1, CTCF, H3K36me3, Rad21
		HSMMtube	H3K27ac, DNase1, H3K9ac, CTCF
		blood (K562)	Rad21, CTCF, H3K36me3
		skin (NHDF-AD)	H3K27ac, H3K9ac, DNase1, H3K4me3, CTCF
		muscle (HSMM)	DNase1, H3K36me3, H3K9ac, H3K4me2, H3K27ac, CTCF
		liver (HepG2)	DNase1, H3K36me3, Rad21, CTCF
		blood (GM12878)	Rad21, CTCF, H3K36me3
		lung (IMR90)	H3K27ac, H3K4me2, H3K18ac, H4K5ac, H4K91ac, CTCF, DNase1
		nervous (NH-A)	H3K9ac, H3K4me2, H3K27ac, DNase1
		skin (NHEK)	DNase1, CTCF, H3K9ac, H3K27ac, H3K4me1

		NHLF	DNase1, CTCF, H3K27ac
		Osteobl	H3K4me2, H3K27ac, CTCF, H3K36me3
		blood (DND-41)	H3K36me3, CTCF
		breast (HMEC)	CTCF, H3K4me2, H3K27ac, DNase1
		cervix (HeLa-S3)	Cjun, DNase1, H3K36me3, CTCF, PolII, Max, TAF1, H3K27ac, H3K4me2, Jund, H3K9ac
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, DNase1, CTCF, Cjun, H3K27ac
		A549	H3K4me3, H3K4me2, CTCF, DNase1, H3K27ac
ENSR00000400379 (open chromatin region)	3	lung (IMR90)	H3K36me3
		skin (NHEK)	H3K36me3
		muscle (HSMM)	DNase1
ENSR00000234322 (promoter flanking region)	1	embryonic stem cell (H1ESC)	H3K36me3, DNase1
		HSMMtube	H3K36me3, DNase1
		blood (K562)	H3K27ac, H3K9ac, Nrsf, FOSL1, DNase1, Ini1, PolII
		skin (NHDF-AD)	H3K4me1, DNase1, H3K4me3
		muscle (HSMM)	DNase1, H3K36me3
		liver (HepG2)	H3K36me3
		blood (GM12878)	H3K36me3
		lung (IMR90)	DNase1, H3K36me3
		nervous (NH-A)	H3K36me3, DNase1
		NHLF	DNase1, H3K27ac
		Osteobl	H3K27ac
		blood (DND-41)	H3K36me3
		cervix (HeLa-S3)	DNase1
		monocytes (Monocytes-CD14+)	H3K36me3
		endothelium (HUVEC)	H3K36me3, Cjun
		A549	H3K4me3, DNase1, H3K36me3
ENSR00000649302 (promoter)	2	embryonic stem cell (H1ESC)	PolII, Rad21, Gabp, TAF7, CTCF, H3K27ac, Yy1, TAF1, H3K4me2, H3K9ac, H3K4me3, DNase1
		HSMMtube	CTCF, H3K9ac, H3K4me2, H3K27ac, H3K4me3, H2AZ, H3K36me3, DNase1
		blood (K562)	H3K79me2, HEY1, H2AZ, TAF1, ELF1, CTCFL, Cmyc, PolII, Gabp, H3K9ac, Rad21, Yy1, Ini1, Egr1, H3K27ac, Max, H3K4me2, CTCF, DNase1, H3K4me3
		skin (NHDF-AD)	H3K9ac, DNase1, H3K4me3, CTCF, H3K4me2, H3K27ac
		muscle (HSMM)	H3K4me3, CTCF, H3K79me2, H2AZ, H3K27ac, H3K4me2, H3K9ac, DNase1, H3K36me3
		liver (HepG2)	Gabp, Yy1, PolII, H3K79me2, TAF1, ELF1, H3K4me1, H2AZ, H3K4me2, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3, H3K36me3, H3K27me3, DNase1
		lung (IMR90)	H3K36me3, H3K4me3, H3K9ac, CTCF, H3K4me2, H3K27ac, DNase1
		blood (GM12878)	TAF1, PolII, BCLAF1, H2AZ, H3K9ac, CTCF, DNase1, Yy1, Gabp, ELF1, H3K79me2, H3K4me3, H3K27ac, H3K4me2
		nervous (NH-A)	DNase1, H3K4me2, H3K4me3, H3K27ac, H3K36me3, CTCF, H3K9ac
		skin (NHEK)	H3K4me2, H3K9ac, H3K4me3, CTCF, DNase1
		NHLF	DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
		Osteobl	H3K36me3, CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K36me3, H3K4me3, H3K9ac, H3K27ac, CTCF, H3K4me2, H3K4me1
		breast (HMEC)	CTCF, H3K4me2, H3K9ac, H3K4me3, H3K27ac, DNase1
		cervix (HeLa-S3)	DNase1, H3K79me2, H3K4me3, Max, TAF1, H3K27ac, H3K4me2, H3K9ac, Ini1, Gabp, PolII, Cmyc, H3K36me3, CTCF
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, CTCF, H3K4me2, H3K27ac, H3K9ac, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, PolII, CTCF, DNase1, Cmyc, H3K27ac, H3K9ac, H3K4me3, Max, H3K4me2
		A549	H3K36me3, H3K27ac, DNase1, CTCF, H3K4me3, H3K4me2, H3K9ac
ENSR00001665325 (open chromatin region)	1	lung (IMR90)	H3K27me3

Variation in RISC binding site

gene	variant(s)	affected transcript(s)	targeting miRNA(s)
NFS1	1	ENST00000374085	hsa-miR-103a-3p
		ENST00000374092	hsa-miR-15a-5p
		ENST00000397425	hsa-miR-15b-5p
		ENST00000440385	hsa-miR-16-5p
		ENST00000480655	hsa-miR-195-5p
		ENST00000498084	hsa-miR-34a-5p
			hsa-miR-424-5p
			hsa-miR-449a
			hsa-miR-497-5p
			hsa-miR-503-5p

Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
CNBD2	upstream gene variant	651	ENST00000349339	NM_080834.2	ENSP00000340954	1
CNBD2	upstream gene variant, downstream gene variant	36	ENST00000489667	NM_001304367.1	ENSP00000480804	3
CNBD2	upstream gene variant	667	ENST00000463258	?	ENSP00000476014	1
CNBD2	upstream gene variant	121	ENST00000622112	?	ENSP00000479333	3
CNBD2	upstream gene variant	667	ENST00000538900	NM_001207076.1	ENSP00000442729	1
CNBD2	upstream gene variant	634	ENST00000373973	?	ENSP00000363084	1
CNBD2	upstream gene variant, downstream gene variant	264	ENST00000614708	?	ENSP00000478833	5
COX7BP2	downstream gene variant, upstream gene variant	413	ENST00000437976	?	?	3
CPNE1	downstream gene variant, upstream gene variant	97	ENST00000414711	?	ENSP00000409955	5
CPNE1	downstream gene variant, upstream gene variant	73	ENST00000483359	?	?	5
CPNE1	downstream gene variant, upstream gene variant	2289	ENST00000498056	?	?	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000317677	NM_003915.5	ENSP00000317257	5
CPNE1	downstream gene variant, upstream gene variant	51	ENST00000412056	?	ENSP00000416962	5
CPNE1	downstream gene variant, upstream gene variant	59	ENST00000397442	?	ENSP00000380584	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000437340	?	ENSP00000415597	4
CPNE1	downstream gene variant, upstream gene variant	295	ENST00000415920	?	ENSP00000396069	4
CPNE1	downstream gene variant, upstream gene variant	52	ENST00000439669	?	ENSP00000409305	5
CPNE1	downstream gene variant, upstream gene variant	57	ENST00000401607	?	ENSP00000386067	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	4
CPNE1	downstream gene variant, upstream gene variant	2	ENST00000430570	?	ENSP00000390626	5
CPNE1	downstream gene variant, upstream gene variant	778	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000498814	?	?	4
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000462352	?	?	4
CPNE1	downstream gene variant, upstream gene variant	48	ENST00000352393	?	ENSP00000336945	5
CPNE1	downstream gene variant, upstream gene variant	679	ENST00000483495	?	?	3
CPNE1	downstream gene variant, upstream gene variant	1639	ENST00000473373	?	?	4
CPNE1	downstream gene variant, upstream gene variant	52	ENST00000440240	?	ENSP00000397638	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000434795	?	ENSP00000409794	5
CPNE1	downstream gene variant, upstream gene variant	71	ENST00000416778	?	ENSP00000389662	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000420363	?	ENSP00000401915	5
CPNE1	downstream gene variant, upstream gene variant	34	ENST00000475146	?	?	6
CPNE1	downstream gene variant, upstream gene variant	45	ENST00000458038	?	ENSP00000390141	5
CPNE1	downstream gene variant, upstream gene variant	376	ENST00000435747	?	ENSP00000412806	5
CPNE1	downstream gene variant, upstream gene variant	33	ENST00000437100	?	ENSP00000391483	5
CPNE1	downstream gene variant, upstream gene variant	2	ENST00000439806	?	ENSP00000387434	5
CPNE1	downstream gene variant, upstream gene variant	207	ENST00000486021	?	?	3

CPNE1	downstream gene variant, upstream gene variant	18	ENST00000414664 ?	ENSP00000404355	5
FER1L4	upstream gene variant	4526	ENST00000426117 ?	?	1
FER1L4	upstream gene variant	4561	ENST00000615531 ?	?	1
FER1L4	upstream gene variant	4493	ENST00000430275 ?	?	1
HIGD1AP16	upstream gene variant, downstream gene variant	560	ENST00000450986 ?	?	4
NFS1	upstream gene variant	2373	ENST00000421540 ?	?	2
NFS1	upstream gene variant	2424	ENST00000306750 ?	ENSP00000304740	2
NFS1	upstream gene variant	2539	ENST00000419569 ?	ENSP00000393482	2
NFS1	downstream gene variant	3230	ENST00000480655 ?	?	3
NFS1	downstream gene variant, upstream gene variant	109	ENST00000541387 NM_001198989.1	ENSP00000440897	5
NFS1	downstream gene variant, upstream gene variant	2539	ENST00000374085 ?	ENSP00000363198	5
NFS1	downstream gene variant	3230	ENST00000498084 ?	?	3
NFS1	downstream gene variant, upstream gene variant	2379	ENST00000397425 ?	ENSP00000380570	5
NFS1	downstream gene variant, upstream gene variant	2373	ENST00000440385 ?	ENSP00000392871	5
NFS1	downstream gene variant, upstream gene variant	2321	ENST00000374092 NM_021100.4	ENSP00000363205	5
NFS1	upstream gene variant	2376	ENST00000456462 ?	ENSP00000397572	2
NFS1	downstream gene variant	554	ENST00000471137 ?	?	2
NFS1	upstream gene variant	2376	ENST00000413203 ?	ENSP00000393669	2
NFS1	downstream gene variant, upstream gene variant	861	ENST00000489163 ?	?	3
PHF20	upstream gene variant, downstream gene variant	2735	ENST00000485604 ?	?	4
PHF20	upstream gene variant, downstream gene variant	492	ENST00000496305 ?	?	3
PHF20	upstream gene variant, downstream gene variant	716	ENST00000473943 ?	?	5
PHF20	upstream gene variant, downstream gene variant	445	ENST00000495338 ?	?	6
PHF20	upstream gene variant, downstream gene variant	2064	ENST00000449988 ?	ENSP00000391915	2
PHF20	upstream gene variant, downstream gene variant	361	ENST00000374000 ?	ENSP00000363112	6
PHF20	upstream gene variant, downstream gene variant	1547	ENST00000480940 ?	?	3
PHF20	upstream gene variant, downstream gene variant	2309	ENST00000420233 ?	ENSP00000413712	4
PHF20	upstream gene variant, downstream gene variant	422	ENST00000486408 ?	?	5
PHF20	upstream gene variant, downstream gene variant	361	ENST00000481202 ?	?	6
PHF20	upstream gene variant, downstream gene variant	309	ENST00000461122 ?	?	6
PHF20	upstream gene variant, downstream gene variant	361	ENST00000339089 ?	ENSP00000341900	6
PHF20	upstream gene variant, downstream gene variant	318	ENST00000374012 NM_016436.4	ENSP00000363124	9
PHF20	upstream gene variant, downstream gene variant	357	ENST00000452270 ?	ENSP00000404455	6
PHF20	upstream gene variant, downstream gene variant	368	ENST00000617560 ?	?	8
PHF20	upstream gene variant, downstream gene variant	347	ENST00000461405 ?	?	11
PHF20	upstream gene variant, downstream gene variant	606	ENST00000439301 ?	ENSP00000410373	5
RBM12	downstream gene variant, upstream gene variant	18	ENST00000374114 NM_001198838.1, NM_006047.5	ENSP00000363228	6

RBM12	downstream gene variant, upstream gene variant	46	ENST00000359646	NM_001198840.1	ENSP00000352668 5
RBM12	upstream gene variant	800	ENST00000435161	?	ENSP00000411692 2
RBM12	downstream gene variant, upstream gene variant	34	ENST00000374104	NM_152838.3	ENSP00000363217 5
RBM12	upstream gene variant	45	ENST00000424458	?	ENSP00000411036 3
RBM12	upstream gene variant	778	ENST00000431148	?	ENSP00000392642 2
RBM39	downstream gene variant, upstream gene variant	624	ENST00000433027	?	ENSP00000407466 6
RBM39	downstream gene variant, upstream gene variant	1250	ENST00000463004	?	? 3
RBM39	downstream gene variant, upstream gene variant	2018	ENST00000496183	?	? 4
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000429968	?	ENSP00000395124 3
RBM39	downstream gene variant, upstream gene variant	2072	ENST00000470563	?	? 5
RBM39	downstream gene variant, upstream gene variant	1252	ENST00000403542	?	ENSP00000385977 3
RBM39	downstream gene variant, upstream gene variant	794	ENST00000498280	?	? 9
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000495293	?	? 5
RBM39	downstream gene variant, upstream gene variant	1221	ENST00000493853	?	? 3
RBM39	downstream gene variant, upstream gene variant	525	ENST00000449489	?	ENSP00000390303 6
RBM39	downstream gene variant, upstream gene variant	2046	ENST00000465158	?	? 5
RBM39	downstream gene variant, upstream gene variant	903	ENST00000397370	?	ENSP00000380527 8
RBM39	downstream gene variant, upstream gene variant	2542	ENST00000374038	?	ENSP00000363150 3
RBM39	upstream gene variant	3407	ENST00000434927	?	ENSP00000393493 1
RBM39	downstream gene variant, upstream gene variant	1567	ENST00000455343	?	ENSP00000395563 7
RBM39	downstream gene variant	333	ENST00000481037	?	? 2
RBM39	downstream gene variant, upstream gene variant	1717	ENST00000416108	?	ENSP00000406900 7
RBM39	downstream gene variant, upstream gene variant	1252	ENST00000338163	?	ENSP00000344581 3
RBM39	downstream gene variant, upstream gene variant	274	ENST00000425184	?	ENSP00000389459 3
RBM39	downstream gene variant, upstream gene variant	97	ENST00000477334	?	? 6
RBM39	downstream gene variant, upstream gene variant	164	ENST00000426951	?	ENSP00000405916 6
RBM39	downstream gene variant, upstream gene variant	694	ENST00000490354	?	? 4
RBM39	downstream gene variant, upstream gene variant	48	ENST00000487604	?	? 6
RBM39	downstream gene variant, upstream gene variant	1363	ENST00000468086	?	? 3
RBM39	downstream gene variant, upstream gene variant	495	ENST00000442447	?	? 6
RBM39	downstream gene variant, upstream gene variant	2391	ENST00000471635	?	? 4
RBM39	downstream gene variant, upstream gene variant	823	ENST00000416529	?	ENSP00000407150 8
RBM39	downstream gene variant, upstream gene variant	87	ENST00000615771	?	? 6
RBM39	downstream gene variant, upstream gene variant	546	ENST00000453310	?	ENSP00000397443 8
RBM39	downstream gene variant, upstream gene variant	736	ENST00000475651	?	? 3
RBM39	downstream gene variant, upstream gene variant	1555	ENST00000463098	?	? 2
RBM39	downstream gene variant, upstream gene variant	1645	ENST00000448303	?	ENSP00000394824 9

RBM39	downstream gene variant, upstream gene variant	862	ENST00000461849 ?	?	3
RBM39	downstream gene variant, upstream gene variant	1967	ENST00000444878 ?	ENSP00000394602	3
RBM39	upstream gene variant	2903	ENST00000412738 ?	ENSP00000397917	1
RBM39	downstream gene variant, upstream gene variant	31	ENST00000492779 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1236	ENST00000476806 ?	ENSP00000483837	5
RBM39	downstream gene variant, upstream gene variant	2018	ENST00000482563 ?	?	4
RBM39	downstream gene variant, upstream gene variant	1251	ENST00000361162 NM_004902.3	ENSP00000354437	3
RBM39	downstream gene variant, upstream gene variant	200	ENST00000461283 ?	?	3
RBM39	downstream gene variant, upstream gene variant	1539	ENST00000528062 NM_001242600.1, NM_001242599.1	ENSP00000436747	3
RBM39	downstream gene variant, upstream gene variant	1080	ENST00000490484 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1129	ENST00000494274 ?	?	6
RBM39	downstream gene variant, upstream gene variant	1251	ENST00000253363 NM_184234.2	ENSP00000253363	3
RN7SKP271	upstream gene variant, downstream gene variant	1031	ENST00000410528 ?	?	4
RNU4-40P	upstream gene variant, downstream gene variant	3161	ENST00000364351 ?	?	5
RNU6-759P	downstream gene variant, upstream gene variant	170	ENST00000516740 ?	?	3
RNU6-937P	downstream gene variant	207	ENST00000384325 ?	?	2
ROMO1	upstream gene variant, downstream gene variant	748	ENST00000336695 ?	ENSP00000338293	4
ROMO1	upstream gene variant, downstream gene variant	744	ENST00000374077 ?	ENSP00000363190	4
ROMO1	upstream gene variant, downstream gene variant	744	ENST00000374078 NM_080748.2	ENSP00000363191	4
ROMO1	upstream gene variant, downstream gene variant	750	ENST00000374072 ?	ENSP00000363185	4
ROMO1	upstream gene variant, downstream gene variant	818	ENST00000397416 ?	ENSP00000380561	4
RP1-309K20.6	downstream gene variant	1589	ENST00000441563 ?	ENSP00000411232	2
RP1-309K20.6	downstream gene variant	1589	ENST00000454607 ?	ENSP00000395201	2
RP1-309K20.6	downstream gene variant	1589	ENST00000541176 ?	ENSP00000443983	2
RPF2P1	upstream gene variant, downstream gene variant	461	ENST00000424137 ?	?	11
SCAND1	downstream gene variant, upstream gene variant	577	ENST00000305978 NM_033630.2	ENSP00000301995	5
SCAND1	downstream gene variant, upstream gene variant	1943	ENST00000615116 NM_016558.3	ENSP00000481289	5
SCAND1	downstream gene variant, upstream gene variant	369	ENST00000373991 ?	ENSP00000363103	5
SPAG4	upstream gene variant, downstream gene variant	3180	ENST00000430878 ?	ENSP00000399231	3
SPAG4	upstream gene variant	1537	ENST00000462896 ?	?	2
SPAG4	upstream gene variant, downstream gene variant	2202	ENST00000498203 ?	?	3
SPAG4	upstream gene variant, downstream gene variant	1537	ENST00000454819 ?	ENSP00000396670	4
SPAG4	upstream gene variant, downstream gene variant	405	ENST00000374273 NM_003116.1	ENSP00000363391	4
SPAG4	upstream gene variant	1393	ENST00000463973 ?	?	2
SPAG4	upstream gene variant, downstream gene variant	1559	ENST00000468248 ?	?	4
Y_RNA	downstream gene variant, upstream gene variant	255	ENST00000362806 ?	?	4

Putative effect on transcript

Intron variant (splice region)

gene	affected transcript	RefSeq id	protein	variant(s)
SCAND1	ENST00000615116	NM_016558.3	ENSP00000481289	1
SCAND1	ENST00000305978	NM_033630.2	ENSP00000301995	1

Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNBD2	ENST00000622112	?	ENSP00000479333	8
CNBD2	ENST00000373973	?	ENSP00000363084	3
CNBD2	ENST00000463258	?	ENSP00000476014	3
CNBD2	ENST00000538900	NM_001207076.1	ENSP00000442729	3
CNBD2	ENST00000349339	NM_080834.2	ENSP00000340954	3
CNBD2	ENST00000489667	NM_001304367.1	ENSP00000480804	1
CPNE1	ENST00000401607	?	ENSP00000386067	10
CPNE1	ENST00000434795	?	ENSP00000409794	9
CPNE1	ENST00000430570	?	ENSP00000390626	10
CPNE1	ENST00000352393	?	ENSP00000336945	10
CPNE1	ENST00000458038	?	ENSP00000390141	9
CPNE1	ENST00000483359	?	?	10
CPNE1	ENST00000414664	?	ENSP00000404355	9
CPNE1	ENST00000414711	?	ENSP00000409955	9
CPNE1	ENST00000415920	?	ENSP00000396069	1
CPNE1	ENST00000483495	?	?	1
CPNE1	ENST00000439669	?	ENSP00000409305	9
CPNE1	ENST00000435747	?	ENSP00000412806	9
CPNE1	ENST00000440240	?	ENSP00000397638	9
CPNE1	ENST00000420363	?	ENSP00000401915	9
CPNE1	ENST00000412056	?	ENSP00000416962	9
CPNE1	ENST00000439806	?	ENSP00000387434	9
CPNE1	ENST00000437340	?	ENSP00000415597	11
CPNE1	ENST00000437100	?	ENSP00000391483	9
CPNE1	ENST00000416778	?	ENSP00000389662	9
CPNE1	ENST00000397442	?	ENSP00000380584	10
CPNE1	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	11
CPNE1	ENST00000475146	?	?	1
CPNE1	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	11
CPNE1	ENST00000317677	NM_003915.5	ENSP00000317257	6
NFS1	ENST00000419569	?	ENSP00000393482	2
NFS1	ENST00000480655	?	?	2
NFS1	ENST00000456462	?	ENSP00000397572	2
NFS1	ENST00000498084	?	?	2

NFS1	ENST00000489163	?	?	1
NFS1	ENST00000374092	NM_021100.4	ENSP00000363205	7
NFS1	ENST00000421540	?	?	2
NFS1	ENST00000374085	?	ENSP00000363198	7
NFS1	ENST00000541387	NM_001198989.1	ENSP00000440897	6
NFS1	ENST00000397425	?	ENSP00000380570	7
NFS1	ENST00000413203	?	ENSP00000393669	5
NFS1	ENST00000440385	?	ENSP00000392871	7
NFS1	ENST00000306750	?	ENSP00000304740	2
PHF20	ENST00000496305	?	?	6
PHF20	ENST00000486408	?	?	7
PHF20	ENST00000439301	?	ENSP00000410373	54
PHF20	ENST00000480940	?	?	6
PHF20	ENST00000495338	?	?	17
PHF20	ENST00000617560	?	?	1
PHF20	ENST00000461405	?	?	16
PHF20	ENST00000452270	?	ENSP00000404455	41
PHF20	ENST00000449988	?	ENSP00000391915	9
PHF20	ENST00000374000	?	ENSP00000363112	70
PHF20	ENST00000481202	?	?	70
PHF20	ENST00000461122	?	?	50
PHF20	ENST00000339089	?	ENSP00000341900	70
PHF20	ENST00000374012	NM_016436.4	ENSP00000363124	81
RBM12	ENST00000374114	NM_001198838.1, NM_006047.5	ENSP00000363228	4
RBM12	ENST00000374104	NM_152838.3	ENSP00000363217	4
RBM12	ENST00000435161	?	ENSP00000411692	5
RBM12	ENST00000424458	?	ENSP00000411036	4
RBM12	ENST00000431148	?	ENSP00000392642	5
RBM12	ENST00000359646	NM_001198840.1	ENSP00000352668	4
RBM39	ENST00000492779	?	?	17
RBM39	ENST00000338163	?	ENSP00000344581	22
RBM39	ENST00000468086	?	?	1
RBM39	ENST00000453310	?	ENSP00000397443	2
RBM39	ENST00000615771	?	?	5
RBM39	ENST00000448303	?	ENSP00000394824	13
RBM39	ENST00000494274	?	?	6
RBM39	ENST00000461283	?	?	21
RBM39	ENST00000490484	?	?	6
RBM39	ENST00000397370	?	ENSP00000380527	2
RBM39	ENST00000434927	?	ENSP00000393493	15
RBM39	ENST00000465158	?	?	3
RBM39	ENST00000412738	?	ENSP00000397917	15

RBM39	ENST00000429968	?	ENSP00000395124	22
RBM39	ENST00000374038	?	ENSP00000363150	15
RBM39	ENST00000361162	NM_004902.3	ENSP00000354437	22
RBM39	ENST00000481037	?	?	13
RBM39	ENST00000475651	?	?	4
RBM39	ENST00000425184	?	ENSP00000389459	13
RBM39	ENST00000416529	?	ENSP00000407150	2
RBM39	ENST00000455343	?	ENSP00000395563	9
RBM39	ENST00000416108	?	ENSP00000406900	9
RBM39	ENST00000253363	NM_184234.2	ENSP00000253363	22
RBM39	ENST00000426951	?	ENSP00000405916	7
RBM39	ENST00000403542	?	ENSP00000385977	22
RBM39	ENST00000477334	?	?	6
RBM39	ENST00000444878	?	ENSP00000394602	22
RBM39	ENST00000487604	?	?	5
RBM39	ENST00000463004	?	?	22
RBM39	ENST00000442447	?	?	6
RBM39	ENST00000496183	?	?	1
RBM39	ENST00000449489	?	ENSP00000390303	6
RBM39	ENST00000482563	?	?	1
RBM39	ENST00000433027	?	ENSP00000407466	6
RBM39	ENST00000528062	NM_001242600.1, NM_001242599.1	ENSP00000436747	22
RBM39	ENST00000471635	?	?	2
RBM39	ENST00000495293	?	?	3
RBM39	ENST00000476806	?	ENSP00000483837	3
RBM39	ENST00000470563	?	?	3
RP1-309K20.6	ENST00000441563	?	ENSP00000411232	13
RP1-309K20.6	ENST00000454607	?	ENSP00000395201	13
RP1-309K20.6	ENST00000541176	?	ENSP00000443983	13
SCAND1	ENST00000373991	?	ENSP00000363103	2

3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CNBD2	ENST00000614708	?	ENSP00000478833	1
NFS1	ENST00000374092	NM_021100.4	ENSP00000363205	1
NFS1	ENST00000374085	?	ENSP00000363198	1
NFS1	ENST00000397425	?	ENSP00000380570	1
NFS1	ENST00000440385	?	ENSP00000392871	1
PHF20	ENST00000374012	NM_016436.4	ENSP00000363124	1
RBM39	ENST00000426951	?	ENSP00000405916	1

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
CPNE1	ENST00000437340	?	ENSP00000415597	1
CPNE1	ENST00000397445	NM_152927.2, NM_152928.2	ENSP00000380587	1
CPNE1	ENST00000397443	NM_152925.2, NM_001198863.1, NM_152926.2	ENSP00000380585	1
RBM12	ENST00000435161	?	ENSP00000411692	1
RBM12	ENST00000431148	?	ENSP00000392642	1
SCAND1	ENST00000373991	?	ENSP00000363103	2
SCAND1	ENST00000615116	NM_016558.3	ENSP00000481289	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
CPNE1	ENST00000483359	?	1
CPNE1	ENST00000483495	?	1
NFS1	ENST00000480655	?	1
NFS1	ENST00000498084	?	1
RBM39	ENST00000492779	?	2
RBM39	ENST00000481037	?	3
RBM39	ENST00000463098	?	1
RBM39	ENST00000477334	?	2
RBM39	ENST00000463004	?	3