

# SNiPacard

## Block annotations

### Block info


genomic range	chr6:26,454,363-26,949,672 <i>el</i>
block size	495,310 bp
variant count	57 variants

### Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.372$ [-9.929 – 5.311]	gene(s) hit or close-by	ABT1 <i>el</i> , BTN1A1 <i>el</i> , BTN2A1 <i>el</i> , BTN3A3 <i>el</i> , CTA-14H9.5 <i>el</i> , GUSBP2 <i>el</i> , HCG11 <i>el</i> , HMGNA4 <i>el</i> , LINC00240 <i>el</i> , POM121L6P <i>el</i> , RP11-457M11.6 <i>el</i> , RP11-457M11.7 <i>el</i> , ZNF322 <i>el</i>
phastCons	$\mu = 0.107$ [0 – 1]	eQTL gene(s)	BTN2A2 <i>el</i> , BTN3A2 <i>el</i> , BTN3A3 <i>el</i> , HIST1H2BD <i>el</i> , HIST1H4H <i>el</i> , HIST1H4J <i>el</i> , PRSS16 <i>el</i> , SCGN <i>el</i> , TRIM38 <i>el</i> , ZNF391 <i>el</i>
GERP++	$\mu = -0.435$ [-5.9 – 3.15]	potentially regulated gene(s)	ABT1 <i>el</i> , BTN1A1 <i>el</i> , BTN3A1 <i>el</i> , BTN3A2 <i>el</i> , BTN3A3 <i>el</i> , HCG11 <i>el</i> , HIST1H2AD <i>el</i> , HIST1H2BD <i>el</i> , HIST1H2BF <i>el</i> , HIST1H3F <i>el</i> , HIST1H4C <i>el</i> , HIST1H4D <i>el</i> , HIST1H4E <i>el</i> , HMGNA4 <i>el</i> , VN1R13P <i>el</i>
CADD score	$\mu = 5.289$ [0.005 – 24.6]	disease gene(s)	-

## Trait annotations

### Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
phenol sulfate	8.78×10 <sup>-7</sup>	Metabolomics GWAS Server	24816252 	5
Lupus erythematosus, systemic	7.66×10 <sup>-6</sup>	dbGaP	pha002867 <b>dbGaP</b>	1
Lupus erythematosus, systemic	1.17×10 <sup>-10</sup>	dbGaP	pha002848 <b>dbGaP</b>	1



## 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)
BTN1A1 <i>el</i>	missense variant	ENST00000244513 <i>el</i>	NM_001732.2	ENSP00000244513	S/P	Tcc/Ccc	?	?	1
BTN1A1 <i>el</i>	missense variant	ENST00000613186 <i>el</i>	?	ENSP00000484707	S/P	Tcc/Ccc	?	?	1
BTN2A1 <i>el</i>	missense variant	ENST00000429381 <i>el</i>	NM_078476.3	ENSP00000416945	3	3			3
BTN2A1 <i>el</i>	missense variant	ENST00000541522 <i>el</i>	NM_001197233.2	ENSP00000443909	5	5			5
BTN2A1 <i>el</i>	missense variant	ENST00000312541 <i>el</i>	NM_007049.4	ENSP00000312158	5	5			5
BTN2A1 <i>el</i>	missense variant	ENST00000469185 <i>el</i>	NM_001197234.2	ENSP00000419043	3	3			3

## Direct effect on regulation

### cis-eQTL



gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
BTN3A2 <i>el</i>	?	ENSG00000186470 <i>el</i>	atrial appendage	1.06×10 <sup>-18</sup> (p-value)	GTEx Portal V6 	49
BTN3A2 <i>el</i>	?	ENSG00000186470 <i>el</i>	transformed fibroblasts	1.95×10 <sup>-29</sup> (p-value)	GTEx Portal V6 	49

ZNF391	e!	?	ENSG00000124613	e!	transformed fibroblasts	1.19×10 <sup>-5</sup> (p-value)	GTEX Portal V6	19
BTN3A2	e!	?	ENSG00000186470	e!	breast	2.31×10 <sup>-16</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	blood	3.20×10 <sup>-54</sup> (p-value)	GTEX Portal V6	49
TRIM38	e!	?	ENSG00000112343	e!	blood	5.60×10 <sup>-6</sup> (p-value)	GTEX Portal V6	2
BTN3A2	e!	?	ENSG00000186470	e!	skeletal muscle	3.00×10 <sup>-43</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	EBV lymphocytes	1.45×10 <sup>-9</sup> (p-value)	GTEX Portal V6	28
BTN3A2	e!	?	ENSG00000186470	e!	unexposed skin	4.12×10 <sup>-14</sup> (p-value)	GTEX Portal V6	49
BTN2A2	e!	?	ENSG00000124508	e!	sun exposed skin	4.82×10 <sup>-6</sup> (p-value)	GTEX Portal V6	11
BTN3A2	e!	?	ENSG00000186470	e!	sun exposed skin	2.65×10 <sup>-35</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	ovary	1.34×10 <sup>-11</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	left ventricle	4.58×10 <sup>-34</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	aorta	5.55×10 <sup>-26</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	subcutaneous adipocytes	1.65×10 <sup>-32</sup> (p-value)	GTEX Portal V6	49
HIST1H4J	e!	?	ENSG00000197238	e!	subcutaneous adipocytes	1.69×10 <sup>-6</sup> (p-value)	GTEX Portal V6	4
BTN3A2	e!	?	ENSG00000186470	e!	caudate basal ganglia	5.23×10 <sup>-6</sup> (p-value)	GTEX Portal V6	28
BTN3A2	e!	?	ENSG00000186470	e!	visceral adipocytes	1.56×10 <sup>-12</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	stomach	1.77×10 <sup>-21</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	cortex	6.44×10 <sup>-8</sup> (p-value)	GTEX Portal V6	31
BTN3A2	e!	?	ENSG00000186470	e!	tibial nerve	4.81×10 <sup>-36</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	pituitary	3.74×10 <sup>-7</sup> (p-value)	GTEX Portal V6	31
BTN3A2	e!	?	ENSG00000186470	e!	esophagus mucosa	1.14×10 <sup>-33</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	testis	6.84×10 <sup>-17</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	hypothalamus	6.80×10 <sup>-8</sup> (p-value)	GTEX Portal V6	31
BTN3A2	e!	?	ENSG00000186470	e!	frontal cortex	1.34×10 <sup>-11</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	uterus	6.66×10 <sup>-12</sup> (p-value)	GTEX Portal V6	38
BTN3A2	e!	?	ENSG00000186470	e!	pancreas	5.39×10 <sup>-18</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	muscularis mucosae	8.36×10 <sup>-32</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	lung	1.27×10 <sup>-42</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	tibial artery	2.40×10 <sup>-39</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	thyroid	2.94×10 <sup>-30</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	transverse colon	7.87×10 <sup>-18</sup> (p-value)	GTEX Portal V6	49
BTN3A2	e!	?	ENSG00000186470	e!	liver	5.30×10 <sup>-9</sup> (p-value)	GTEX Portal V6	47
BTN3A2	e!	?	ENSG00000186470	e!	nucleus accumbens	1.93×10 <sup>-7</sup> (p-value)	GTEX Portal V6	31
BTN3A2	e!	?	ENSG00000186470	e!	spleen	2.37×10 <sup>-10</sup> (p-value)	GTEX Portal V6	47

BTN3A2	e!	?	ENSG00000186470	e!	coronary artery	6.37×10 <sup>-16</sup> (p-value)	GTEEx Portal V6		49	
BTN3A2	e!	?	ENSG00000186470	e!	vagina	3.81×10 <sup>-15</sup> (p-value)	GTEEx Portal V6		49	
BTN3A2	e!	?	ENSG00000186470	e!	gastroesophageal junction	1.27×10 <sup>-20</sup> (p-value)	GTEEx Portal V6		49	
BTN3A2	e!	?	ENSG00000186470	e!	adrenal gland	2.01×10 <sup>-11</sup> (p-value)	GTEEx Portal V6		49	
BTN3A2	e!	?	ENSG00000186470	e!	cerebellum	1.03×10 <sup>-8</sup> (p-value)	GTEEx Portal V6		47	
PRSS16	e!	?	ENSG00000112812	e!	cerebellum	2.12×10 <sup>-7</sup> (p-value)	GTEEx Portal V6		18	
SCGN	e!	ENST00000334979	e!	ILMN_1789648	e!	adipocyte	9.10×10 <sup>-6</sup> (p-value)	MuTHER consortium		4
SCGN	e!	ENST00000612225	e!							
SCGN	e!	ENST00000377961	e!							
BTN2A2	e!	ENST00000471116	e!	ILMN_2256894	e!	blood	5.64×10 <sup>-5</sup> (p-value)	Westra et al.		3
BTN2A2	e!	ENST00000356709	e!							
BTN2A2	e!	ENST00000469230	e!							
BTN2A2	e!	ENST00000482636	e!							
BTN2A2	e!	ENST00000494184	e!							
BTN2A2	e!	ENST00000416795	e!							
BTN2A2	e!	ENST00000432533	e!							
BTN2A2	e!	ENST00000493275	e!							
HIST1H4H	e!	ENST00000377727	e!	ILMN_1751120	e!	blood	8.40×10 <sup>-6</sup> (p-value)	Westra et al.		3
BTN3A3	e!	?	ENSG00000111801	e!	blood	4.34×10 <sup>-4</sup> (q-value)	SeeQTL DB (HapMap)		2	
BTN3A2	e!	?	ENSG00000186470	e!	blood	4.56×10 <sup>-12</sup> (q-value)	SeeQTL DB (HapMap)		2	
HIST1H2BD	e!	ENST00000289316	e!	ILMN_1651496	e!	monocyte	2.65×10 <sup>-12</sup> (p-value)	Zeller et al.		1
BTN3A2	e!	ENST00000396934	e!	ILMN_1676528	e!	monocyte	8.02×10 <sup>-83</sup> (p-value)	Zeller et al.		2
BTN3A2	e!	ENST00000508906	e!							
BTN3A2	e!	ENST00000356386	e!							
?	?	?	ILMN_1820787	e!	monocyte	2.91×10 <sup>-58</sup> (p-value)	Zeller et al.		2	
?	?	?	ILMN_1883997	e!	monocyte	5.68×10 <sup>-16</sup> (p-value)	Zeller et al.		2	
BTN3A2	e!	ENST00000524459	e!	ILMN_1700067	e!	monocyte	5.72×10 <sup>-65</sup> (p-value)	Zeller et al.		2
BTN3A2	e!	ENST00000528541	e!							
BTN3A3	e!	ENST00000467524	e!							
BTN3A3	e!	ENST00000482451	e!							
BTN3A3	e!	ENST00000494393	e!							
BTN3A2	e!	ENST00000527417	e!							
BTN3A3	e!	ENST00000244519	e!							
BTN3A2	e!	ENST00000528222	e!							
BTN3A3	e!	ENST00000474790	e!							
BTN3A2	e!	ENST00000377708	e!							
BTN3A3	e!	ENST00000496719	e!							
BTN3A2	e!	ENST00000396948	e!							
BTN3A2	e!	ENST00000527422	e!							
BTN3A2	e!	ENST00000356386	e!							
BTN3A2	e!	ENST00000524682	e!	209846_s_at	e!	blood	7.30×10 <sup>-22</sup> (p-value)	Dixon et al.		1
BTN3A2	e!	ENST00000604202	e!							
BTN3A2	e!	ENST00000396934	e!							
BTN3A2	e!	ENST00000532294	e!							
BTN3A2	e!	ENST00000508906	e!							
BTN3A2	e!	ENST00000377708	e!							

BTN3A2	e!	ENST00000527422	e!						
BTN3A2	e!	ENST00000531055	e!						
BTN3A2	e!	ENST00000396948	e!						
BTN3A2	e!	ENST00000356386	e!						
BTN3A2	e!	ENST00000396934	e!	212613_at	e!	blood	1.40×10 <sup>-19</sup> (p-value)	Dixon et al. 	1
BTN3A2	e!	ENST00000508906	e!						
BTN3A2	e!	ENST00000356386	e!						
BTN3A3	e!	ENST00000244519	e!	204820_s_at	e!	blood	3.50×10 <sup>-21</sup> (p-value)	Dixon et al. 	1
BTN3A2	e!	ENST00000396934	e!						
BTN3A2	e!	ENST00000508906	e!						
BTN3A2	e!	ENST00000356386	e!						
BTN3A3	e!	ENST00000480110	e!						
BTN2A2	e!	ENST00000471116	e!	ILMN_2256894	e!	b-cell	1.99×10 <sup>-5</sup> (p-value)	Fairfax et al. 	1
BTN2A2	e!	ENST00000356709	e!						
BTN2A2	e!	ENST00000494184	e!						
BTN2A2	e!	ENST00000482636	e!						
BTN2A2	e!	ENST00000469230	e!						
BTN2A2	e!	ENST00000416795	e!						
BTN2A2	e!	ENST00000432533	e!						
BTN2A2	e!	ENST00000493275	e!						

### trans-eQTL

gene	transcript	probe	chromosome	tissue	min(statistic) (type)	source	variant(s)
?	?	ILMN_1671054	chr6	monocyte	9.58×10 <sup>-22</sup> (p-value)	Zeller et al. 	2
?	?	ILMN_1752592	chr6	monocyte	1.68×10 <sup>-13</sup> (p-value)	Zeller et al. 	2

### Putative effect on regulation

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

SNiPA enhancer id	variant(s)	associated SNiPA promoter id	associated gene(s)
ENCE00000442938	1	ENCP00000047667	BTN3A2
		ENCP00000047679	BTN1A1
ENCE00000442976	1	ENCP00000047676	BTN3A3
ENCE00000442878	1	ENCP00000047649	HIST1H2AD
			HIST1H2BF
		ENCP00000047646	HIST1H4D
		ENCP00000047671	BTN3A1
		ENCP00000047680	HCG11
		ENCP00000047665	BTN3A2
		ENCP00000047640	HIST1H4C
		ENCP00000047644	HIST1H2BD
		ENCP00000047681	HMGN4
		ENCP00000047658	HIST1H3F
		ENCP00000047682	ABT1
		ENCP00000047674	BTN3A3
		ENCP00000047651	HIST1H4E
ENCE00000443031	1	ENCP00000047691	VN1R13P

### Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001494264	2	monocytes (Monocytes-CD14+)	H3K36me3

(enhancer)		blood (DND-41)	H3K36me3
		skin (NHDF-AD)	H3K4me1, DNase1
ENSR00001214106 <i>e!</i>	1	monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K27ac
(enhancer)		endothelium (HUVEC)	H3K27me3
		liver (HepG2)	H3K4me1
		blood (K562)	PolII, Ini1, DNase1, PU1, H2AZ, H3K4me1
ENSR00001214131 <i>e!</i>	1	embryonic stem cell (H1ESC)	DNase1, H3K27ac, PolII, Rad21, TAF7, Yy1, TAF1, H3K4me2, H3K4me3
(promoter flanking region)		HSMMtube	H3K4me2, DNase1, H2AZ
		blood (K562)	PolII, Ini1, Brg1, DNase1, PolIII, THAP1, H3K27ac, Max, H3K9ac, Cmyc, H2AZ, Nrsf, H3K4me2, H3K4me3
		skin (NHDF-AD)	H3K4me3, DNase1, H3K4me2
		muscle (HSMM)	DNase1, H3K4me2
		liver (HepG2)	Gabp, PolII, H2AZ, H3K4me2, H3K9ac, Cmyc, H3K4me3, H3K27me3, DNase1
		lung (IMR90)	H3K4me2, H3K4me3, DNase1
		blood (GM12878)	PolII, MEF2A, H2AZ, DNase1, Yy1, PolIII, H3K4me3, H3K27ac, H3K4me2, H3K9ac
		nervous (NH-A)	DNase1, H3K4me2, H3K9ac
		skin (NHEK)	DNase1, H3K4me2
		NHLF	H3K4me3, DNase1
		Osteobl	H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, H3K4me2, H3K27ac, H3K9ac, H3K4me3
		breast (HMEC)	DNase1, H3K4me3, H3K4me2
		cervix (HeLa-S3)	DNase1, Max, H3K4me3, Gabp, H3K27ac, H3K4me2, H3K9ac, JunD
		monocytes (Monocytes-CD14+)	DNase1, H3K27me3, H3K4me3
		endothelium (HUVEC)	Cjun, H3K4me2, PolII, DNase1
		A549	H3K9ac, H3K4me2, H3K4me3, DNase1
ENSR00001494272 <i>e!</i>	1	embryonic stem cell (H1ESC)	Rad21, CTCF, H3K4me2, H3K4me3, DNase1
(promoter flanking region)		HSMMtube	DNase1, H3K27me3, H2AZ, H3K4me2, CTCF
		blood (K562)	PolII, Ini1, DNase1, PolIII, Egr1, H3K27ac, Max, Rad21, H3K9ac, H2AZ, H3K4me2, CTCF, H3K4me3
		skin (NHDF-AD)	DNase1, H3K9ac, H3K4me2, H3K4me3, CTCF
		muscle (HSMM)	H2AZ, CTCF, H3K4me3, H3K4me2, DNase1
		liver (HepG2)	DNase1, H3K27me3, Rad21, H2AZ, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF, Cmyc, H3K4me3
		lung (IMR90)	DNase1, H3K4me3, CTCF
		blood (GM12878)	H2AZ, DNase1, Yy1, Rad21, H3K4me3, H3K27ac, H3K4me2, CTCF, H3K9ac
		nervous (NH-A)	DNase1, H3K4me3, H3K4me2, CTCF, H3K9ac
		skin (NHEK)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		NHLF	DNase1, H3K4me3, CTCF
		Osteobl	CTCF, H3K4me2, H3K4me3, H3K27ac, H2AZ
		blood (DND-41)	H3K4me1, CTCF, H3K4me3
		breast (HMEC)	CTCF, H3K4me3, DNase1
		cervix (HeLa-S3)	PolII, H3K9ac, H3K4me2, H3K27ac, H3K4me3, CTCF, DNase1
		monocytes (Monocytes-CD14+)	DNase1, CTCF, H3K4me2, H3K4me3
		endothelium (HUVEC)	H3K36me3, H3K4me2, H3K4me3, CTCF, DNase1, Cmyc
		A549	DNase1, CTCF, H3K9ac, H3K4me2, H3K4me3
ENSR00001214144 <i>e!</i>	2	HSMMtube	H3K27me3
(CTCF binding site)		Osteobl	H3K27me3
		blood (DND-41)	H3K27me3
		blood (K562)	CTCF, H2AZ
		cervix (HeLa-S3)	CTCF
		monocytes (Monocytes-CD14+)	H3K27me3
		endothelium (HUVEC)	H3K27me3
		lung (IMR90)	H3K27me3
		nervous (NH-A)	H3K27me3
ENSR00001702766 <i>e!</i>	1	embryonic stem cell (H1ESC)	TAF1, H3K4me2, SP1, H3K4me3, DNase1, HDAC2, CTCF, H3K27ac, PolII, Rad21, Gabp, TAF7, H3K9ac
(CTCF binding site)		HSMMtube	CTCF, H2AZ, DNase1
		blood (K562)	ELF1, Cfos, CTCFL, PolII, H3K4me2, CTCF, DNase1, H3K4me3, TAF1, H2AZ, H3K27ac, Max, Rad21, H3K9ac, Gabp, Cmyc, HEY1
		skin (NHDF-AD)	CTCF, H3K4me3, DNase1, H3K9ac, H3K4me2
		muscle (HSMM)	H2AZ, CTCF, H3K4me3
		liver (HepG2)	DNase1, Gabp, H3K4me3, PolII, TAF1, ELF1, H3K4me2, H3K9ac, H3K27ac, CTCF
		lung (IMR90)	CTCF, DNase1, H3K4me2, H3K4me3
		blood (GM12878)	DNase1, Gabp, ELF1, Rad21, H3K4me3, H3K27ac, Cfos, CTCF, H3K9ac, PolII, H2AZ
		nervous (NH-A)	DNase1
		skin (NHEK)	DNase1, H3K9ac, H3K4me3, CTCF

ENSR00001214204	1	embryonic stem cell (H1ESC)	DNase1, H3K4me3, H3K27ac, PolII, Rad21, Gabp, TAF7, H3K9ac, CTCF, HDAC2, TAF1, H3K4me2, SP1
NHLF			H3K9ac, H3K27ac, CTCF, H3K4me3, DNase1
Osteobl			CTCF, H3K4me2, H3K4me3
blood (DND-41)			CTCF, H3K27ac, H3K9ac
breast (HMEC)			H3K27ac, H3K4me3, H3K4me2, CTCF
cervix (HeLa-S3)			H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII, CTCF, DNase1
monocytes (Monocytes-CD14+)			CTCF, H3K9ac, H3K4me3
endothelium (HUVEC)			H3K4me3, PolII, CTCF, DNase1
A549			H3K27ac, DNase1, CTCF, H3K4me2, H3K4me3
HSMMtube			H2AZ, CTCF, DNase1
blood (K562)			H3K27ac, Max, Rad21, H3K9ac, Gabp, Cmyc, HEY1, H2AZ, TAF1, ELF1, Cfos, CTCFL, PolII, H3K4me2, CTCF, DNase1, H3K4me3
skin (NHDF-AD)			H3K4me2, H3K9ac, DNase1, CTCF, H3K4me3
muscle (HSMM)			H2AZ, CTCF, H3K4me3
liver (HepG2)			DNase1, Gabp, H3K4me3, FOXA2, PolII, TAF1, ELF1, H3K4me2, FOXA1, H3K9ac, H3K27ac, CTCF
blood (GM12878)			CTCF, H3K9ac, Cfos, H3K27ac, PolII, H2AZ, DNase1, Gabp, ELF1, Rad21, H3K4me3
lung (IMR90)			H3K4me3, CTCF, H3K4me2, DNase1
nervous (NH-A)			DNase1
skin (NHEK)			DNase1, H3K9ac, H3K4me3, CTCF
NHLF			DNase1, H3K4me3, CTCF, H3K9ac, H3K27ac
Osteobl			CTCF, H3K4me2, H3K4me3
blood (DND-41)			CTCF, H3K27ac, H3K9ac
breast (HMEC)			H3K27ac, H3K4me3, H3K4me2, CTCF
cervix (HeLa-S3)			DNase1, CTCF, H3K9ac, H3K4me2, H3K27ac, TAF1, H3K4me3, Gabp, PolII
monocytes (Monocytes-CD14+)			CTCF, H3K9ac, H3K4me3
endothelium (HUVEC)			H3K4me3, PolII, CTCF, DNase1
A549			H3K27ac, DNase1, CTCF, H3K4me2, H3K4me3

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ABT1 <i>el</i>	upstream gene variant, downstream gene variant	2952	ENST00000274849 <i>el</i>	NM_013375.3	ENSP00000274849 <i>el</i>	3
BTN1A1 <i>el</i>	upstream gene variant, downstream gene variant	1094	ENST00000244513 <i>el</i>	NM_001732.2	ENSP00000244513 <i>el</i>	3
BTN1A1 <i>el</i>	upstream gene variant, downstream gene variant	1581	ENST00000613186 <i>el</i> ?		ENSP00000484707 <i>el</i>	3
BTN2A1 <i>el</i>	upstream gene variant	2079	ENST00000312541 <i>el</i>	NM_007049.4	ENSP00000312158 <i>el</i>	4
BTN2A1 <i>el</i>	upstream gene variant	2115	ENST00000429381 <i>el</i>	NM_078476.3	ENSP00000416945 <i>el</i>	4
BTN2A1 <i>el</i>	upstream gene variant	2058	ENST00000541522 <i>el</i>	NM_001197233.2	ENSP00000443909 <i>el</i>	4
BTN2A1 <i>el</i>	upstream gene variant	2076	ENST00000377600 <i>el</i> ?		ENSP00000366825 <i>el</i>	4
BTN2A1 <i>el</i>	upstream gene variant, downstream gene variant	1405	ENST00000480218 <i>el</i> ?		ENSP00000418936 <i>el</i>	6
BTN2A1 <i>el</i>	upstream gene variant, downstream gene variant	93	ENST00000493173 <i>el</i> ?		ENSP00000420447 <i>el</i>	8
BTN2A1 <i>el</i>	upstream gene variant, downstream gene variant	1403	ENST00000469185 <i>el</i>	NM_001197234.2	ENSP00000419043 <i>el</i>	5
BTN3A3 <i>el</i>	downstream gene variant	1320	ENST00000361232 <i>el</i>	NM_197974.2	ENSP00000355238 <i>el</i>	4
BTN3A3 <i>el</i>	downstream gene variant	720	ENST00000244519 <i>el</i>	NM_006994.4	ENSP00000244519 <i>el</i>	4
BTN3A3 <i>el</i>	downstream gene variant	721	ENST00000480110 <i>el</i> ?		?	4
BTN3A3 <i>el</i>	downstream gene variant	2218	ENST00000490254 <i>el</i>	NM_001242803.1	ENSP00000419736 <i>el</i>	4
BTN3A3 <i>el</i>	downstream gene variant	2220	ENST00000497681 <i>el</i> ?		?	4
BTN3A3 <i>el</i>	downstream gene variant	1974	ENST00000483179 <i>el</i> ?		?	4
BTN3A3 <i>el</i>	downstream gene variant	3921	ENST00000477388 <i>el</i> ?		?	2
CTA-14H9.5 <i>el</i>	downstream gene variant	2258	ENST00000567732 <i>el</i> ?		?	2
GUSBP2 <i>el</i>	upstream gene variant	24	ENST00000479900 <i>el</i> ?		?	2
GUSBP2 <i>el</i>	downstream gene variant	2834	ENST00000492832 <i>el</i> ?		?	1
GUSBP2 <i>el</i>	downstream gene variant, upstream gene	2183	ENST00000463434 <i>el</i> ?		?	2

	variant					
HCG11 <i>e!</i>	downstream gene variant	3083	ENST00000411553 <i>e!</i> ?	?		2
HMGN4 <i>e!</i>	downstream gene variant	2730	ENST00000377575 <i>e!</i> NM_006353.2	ENSP00000366798 <i>e!</i>		2
LINC00240 <i>e!</i>	upstream gene variant	415	ENST00000606878 <i>e!</i> ?	?		2
LINC00240 <i>e!</i>	upstream gene variant, downstream gene variant	414	ENST00000607607 <i>e!</i> ?	?		3
POM121L6P <i>e!</i>	downstream gene variant	4071	ENST00000441186 <i>e!</i> ?	?		1
RP11-457M11.6 <i>e!</i>	downstream gene variant	2142	ENST00000602994 <i>e!</i> ?	?		1
RP11-457M11.7 <i>e!</i>	upstream gene variant, downstream gene variant	3100	ENST00000616994 <i>e!</i> ?	?		2
RP11-457M11.7 <i>e!</i>	upstream gene variant, downstream gene variant	3100	ENST00000611708 <i>e!</i> ?	?		2
ZNF322 <i>e!</i>	downstream gene variant	910	ENST00000622479 <i>e!</i> NM_001242797.1	ENSP00000482607 <i>e!</i>		2
ZNF322 <i>e!</i>	downstream gene variant	900	ENST00000415922 <i>e!</i> NM_024639.4	ENSP00000418897 <i>e!</i>		2
ZNF322 <i>e!</i>	downstream gene variant	2808	ENST00000471278 <i>e!</i> ?	ENSP00000419728 <i>e!</i>		2
ZNF322 <i>e!</i>	downstream gene variant	4993	ENST00000480036 <i>e!</i> ?	ENSP00000420301 <i>e!</i>		1
ZNF322 <i>e!</i>	downstream gene variant	910	ENST00000607204 <i>e!</i> NM_001242798.1	ENSP00000483223 <i>e!</i>		2
ZNF322 <i>e!</i>	downstream gene variant	910	ENST00000456172 <i>e!</i> NM_001242799.1	ENSP00000478899 <i>e!</i>		2

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
BTN2A1 <i>e!</i>	ENST00000377600 <i>e!</i>	?	ENSP00000366825 <i>e!</i>	4
BTN2A1 <i>e!</i>	ENST00000312541 <i>e!</i>	NM_007049.4	ENSP00000312158 <i>e!</i>	2
BTN2A1 <i>e!</i>	ENST00000429381 <i>e!</i>	NM_078476.3	ENSP00000416945 <i>e!</i>	4
BTN2A1 <i>e!</i>	ENST00000469185 <i>e!</i>	NM_001197234.2	ENSP00000419043 <i>e!</i>	4
BTN2A1 <i>e!</i>	ENST00000541522 <i>e!</i>	NM_001197233.2	ENSP00000443909 <i>e!</i>	2
BTN2A1 <i>e!</i>	ENST00000480218 <i>e!</i>	?	ENSP00000418936 <i>e!</i>	2
BTN2A1 <i>e!</i>	ENST00000493173 <i>e!</i>	?	ENSP00000420447 <i>e!</i>	2
GUSBP2 <i>e!</i>	ENST00000479900 <i>e!</i>	?	?	9
GUSBP2 <i>e!</i>	ENST00000463434 <i>e!</i>	?	?	3
GUSBP2 <i>e!</i>	ENST00000492832 <i>e!</i>	?	?	1
LINC00240 <i>e!</i>	ENST00000607607 <i>e!</i>	?	?	3
LINC00240 <i>e!</i>	ENST00000606878 <i>e!</i>	?	?	5
RP11-457M11.7 <i>e!</i>	ENST00000611708 <i>e!</i>	?	?	1
ZNF322 <i>e!</i>	ENST00000415922 <i>e!</i>	NM_024639.4	ENSP00000418897 <i>e!</i>	1
ZNF322 <i>e!</i>	ENST00000622479 <i>e!</i>	NM_001242797.1	ENSP00000482607 <i>e!</i>	1
ZNF322 <i>e!</i>	ENST00000465674 <i>e!</i>	?	?	1
ZNF322 <i>e!</i>	ENST00000471278 <i>e!</i>	?	ENSP00000419728 <i>e!</i>	1
ZNF322 <i>e!</i>	ENST00000461899 <i>e!</i>	?	?	1
ZNF322 <i>e!</i>	ENST00000607204 <i>e!</i>	NM_001242798.1	ENSP00000483223 <i>e!</i>	1
ZNF322 <i>e!</i>	ENST00000480036 <i>e!</i>	?	ENSP00000420301 <i>e!</i>	1
ZNF322 <i>e!</i>	ENST00000456172 <i>e!</i>	NM_001242799.1	ENSP00000478899 <i>e!</i>	1

#### 3'-UTR variant

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ABT1 <i>e!</i>	ENST00000274849 <i>e!</i>	NM_013375.3	ENSP00000274849 <i>e!</i>	1
BTN2A1 <i>e!</i>	ENST00000377600 <i>e!</i>	?	ENSP00000366825 <i>e!</i>	5
BTN2A1 <i>e!</i>	ENST00000429381 <i>e!</i>	NM_078476.3	ENSP00000416945 <i>e!</i>	2

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
LINC00240 <i>e!</i>	ENST00000607607 <i>e!</i>	?	1
RP11-457M11.7 <i>e!</i>	ENST00000616994 <i>e!</i>	?	1



