

Block annotations

Block info


genomic range	chr15:60,687,152-60,798,590 <i>e!</i>
block size	111,439 bp
variant count	29 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	$\mu = -0.490$ [-3.197 – 1.559]	gene(s) hit or close-by	ANXA2 <i>e!</i> , ICE2 <i>e!</i> , RORA <i>e!</i> , RORA-AS1 <i>e!</i> , RP11-745A24.1 <i>e!</i> , RP11-745A24.2 <i>e!</i> , RP11-745A24.3 <i>e!</i>
phastCons	$\mu = 0.100$ [0 – 0.992]	eQTL gene(s)	ICE2 <i>e!</i>
GERP++	$\mu = -1.141$ [-7.15 – 3.05]	potentially regulated gene(s)	-
CADD score	$\mu = 3.973$ [0.01 – 13.59]	disease gene(s)	-

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
ICE2 <i>e!</i>	ENST00000261520 <i>e!</i>	ILMN_1666610 <i>e!</i>	blood	4.67×10 <sup>-5</sup> (p-value)	Westra et al. 	3
ICE2 <i>e!</i>	ENST00000558181 <i>e!</i>					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001452708 <i>e!</i> (promoter)	3	embryonic stem cell (H1ESC)	DNase1, Rad21, Jund, Nanog, Yy1, TAF1, H3K4me2, SP1, H3K4me3, PolII, H3K27me3, H3K36me3
		HSMMtube	H3K79me2, H3K4me1, H3K9ac, H3K4me2, DNase1, H3K36me3, H3K4me3, H3K27ac
		blood (K562)	Egr1, H3K27ac, Max, E2F6, H3K9ac, Cmyc, H3K79me2, HEY1, H2AZ, Cjun, PolII, H3K4me2, H3K36me3, DNase1, H3K4me3
		skin (NHDF-AD)	H3K27ac, H3K4me2, H3K9ac, DNase1, H3K4me3, CTCF, H3K4me1, H3K36me3
		muscle (HSMM)	H3K9ac, H3K36me3, DNase1, H3K4me2, H3K27ac, H3K79me2, CTCF, H4K20me1, H3K4me3
		liver (HepG2)	DNase1, FOSL2, Jund, H3K4me3, PolII, H3K79me2, H3K4me1, H3K4me2, FOXA1, H3K9ac, H3K27ac, Cmyc, H3K36me3
		lung (IMR90)	DNase1, H4K20me1, H3K79me2, H3K27ac, H3K18ac, H3K4me2, H3K36me3, H3K4me3, H3K9ac, H4K91ac, H3K56ac, CTCF
		blood (GM12878)	BATF, H2AZ, H3K4me1, DNase1, H3K79me2, H3K4me3, H3K4me2, H3K9ac, H3K36me3, PolII
		nervous (NH-A)	DNase1, H3K36me3, H3K27ac, H3K4me3, H3K4me2, H3K4me1, H4K20me1, H3K9ac
		skin (NHEK)	DNase1, H3K36me3, H3K4me3, H3K4me2, PolII, H3K9ac, H4K20me1, H3K27ac, H3K4me1, CTCF
		NHLF	DNase1, H3K4me3, H3K4me1, H3K9ac, H3K27ac, H3K36me3
		Osteobl	H3K36me3, H3K27ac, H3K4me3, H3K4me2
		blood (DND-41)	H3K27me3
		breast (HMEC)	DNase1, H3K27ac, H3K4me3, H3K9ac, H3K4me2, H3K36me3
		cervix (HeLa-S3)	H3K4me3, H3K79me2, PolII, Cmyc, CTCF, H3K36me3, Ini1, H3K9ac, Jund, H3K4me2, H3K27ac, H3K4me1, TAF1, Max, DNase1
		monocytes (Monocytes-CD14+)	DNase1, H3K4me1, H3K4me2, H3K27ac, H3K9ac, H4K20me1, H3K36me3, H3K4me3
		endothelium (HUVEC)	H3K36me3, Cmyc, DNase1, PolII, H3K27ac, H3K4me1, H3K9ac, H3K4me3, H3K4me2, Cjun
A549	H3K4me3, H3K4me2, H3K9ac, DNase1, H3K27ac, H3K36me3		
ENSR00000409015 <i>e!</i> (promoter flanking region)	1	cervix (HeLa-S3)	Jund, DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3
ENSR00000237536 <i>e!</i>	2	skin (NHDF-AD)	DNase1
		embryonic stem cell (H1ESC)	DNase1, H3K36me3

(promoter flanking region)	HSMMtube	DNase1, H3K27ac	
	blood (K562)	DNase1, H3K36me3	
	skin (NHDF-AD)	DNase1	
	muscle (HSMM)	H3K27ac, H3K9ac, H3K36me3, DNase1	
	liver (HepG2)	H3K4me1	
	lung (IMR90)	DNase1, H3K36me3	
	blood (GM12878)	H3K36me3	
	nervous (NH-A)	DNase1	
	skin (NHEK)	H3K36me3, H3K27ac, H3K4me1, DNase1	
	NHLF	DNase1	
	Osteobl	H3K27ac, H3K4me2, H3K36me3	
	blood (DND-41)	H3K36me3	
	breast (HMEC)	H3K27ac	
	cervix (HeLa-S3)	H3K36me3	
	monocytes (Monocytes-CD14+)	H3K4me1, H3K36me3	
	endothelium (HUVEC)	H3K4me2, Cjun	
	A549	H3K36me3	
	ENSR00001628076 <i>e!</i> 1	monocytes (Monocytes-CD14+)	H3K36me3
	(enhancer)	A549	H3K4me3, H3K36me3

### Variation proximal to gene

gene	variant type	min(distance)	transcript	RefSeq id	protein	variant(s)
ANXA2 <i>e!</i>	upstream gene variant	3802	ENST00000558986 <i>e!</i> ?		ENSP00000453739 <i>e!</i>	1
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000557906 <i>e!</i> ?		ENSP00000452895 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	732	ENST00000558998 <i>e!</i> ?		ENSP00000453925 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	653	ENST00000451270 <i>e!</i> NM_004039.2		ENSP00000387545 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	3826	ENST00000559350 <i>e!</i> ?		ENSP00000453663 <i>e!</i>	1
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000559370 <i>e!</i> ?		ENSP00000453566 <i>e!</i>	2
ANXA2 <i>e!</i>	downstream gene variant, upstream gene variant	692	ENST00000560936 <i>e!</i> ?		?	3
ANXA2 <i>e!</i>	upstream gene variant	723	ENST00000557986 <i>e!</i> ?		ENSP00000453742 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	3656	ENST00000560389 <i>e!</i> ?		ENSP00000453579 <i>e!</i>	1
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000396024 <i>e!</i> NM_001136015.2		ENSP00000379342 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	458	ENST00000560367 <i>e!</i> ?		ENSP00000453556 <i>e!</i>	3
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000561445 <i>e!</i> ?		?	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000559113 <i>e!</i> ?		ENSP00000453869 <i>e!</i>	2
ANXA2 <i>e!</i>	downstream gene variant, upstream gene variant	708	ENST00000560495 <i>e!</i> ?		?	3
ANXA2 <i>e!</i>	upstream gene variant	718	ENST00000559467 <i>e!</i> ?		ENSP00000453000 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000561022 <i>e!</i> ?		ENSP00000452876 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	708	ENST00000559559 <i>e!</i> ?		?	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000558558 <i>e!</i> ?		ENSP00000452981 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000421017 <i>e!</i> NM_001002857.1		ENSP00000411352 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	722	ENST00000559780 <i>e!</i> ?		ENSP00000453770 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	663	ENST00000558985 <i>e!</i> ?		ENSP00000453039 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000557937 <i>e!</i> ?		?	2
ANXA2 <i>e!</i>	upstream gene variant	711	ENST00000332680 <i>e!</i> NM_001002858.2		ENSP00000346032 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	710	ENST00000560468 <i>e!</i> ?		ENSP00000452858 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	695	ENST00000559725 <i>e!</i> ?		ENSP00000453174 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	708	ENST00000558169 <i>e!</i> ?		ENSP00000453945 <i>e!</i>	2
ANXA2 <i>e!</i>	upstream gene variant	728	ENST00000560165 <i>e!</i> ?		ENSP00000452921 <i>e!</i>	2

ANXA2 <i>el</i>	upstream gene variant	2875	ENST00000559818 <i>el</i> ?	ENSP00000453859 <i>el</i>	1
ANXA2 <i>el</i>	upstream gene variant	710	ENST00000560546 <i>el</i> ?	?	2
ANXA2 <i>el</i>	upstream gene variant	712	ENST00000559956 <i>el</i> ?	ENSP00000453694 <i>el</i>	2
ANXA2 <i>el</i>	upstream gene variant	710	ENST00000560466 <i>el</i> ?	ENSP00000453841 <i>el</i>	2
ANXA2 <i>el</i>	upstream gene variant	722	ENST00000558503 <i>el</i> ?	ENSP00000453356 <i>el</i>	2
ANXA2 <i>el</i>	upstream gene variant	710	ENST00000558132 <i>el</i> ?	ENSP00000454096 <i>el</i>	2
ANXA2 <i>el</i>	upstream gene variant	1172	ENST00000557904 <i>el</i> ?	ENSP00000452958 <i>el</i>	1
ICE2 <i>el</i>	downstream gene variant	1253	ENST00000558512 <i>el</i> ?	ENSP00000452714 <i>el</i>	1
ICE2 <i>el</i>	downstream gene variant	4302	ENST00000560668 <i>el</i> ?	ENSP00000453303 <i>el</i>	1
ICE2 <i>el</i>	downstream gene variant, upstream gene variant	1773	ENST00000561328 <i>el</i> ?	?	4
ICE2 <i>el</i>	downstream gene variant	603	ENST00000561124 <i>el</i> ?	?	2
ICE2 <i>el</i>	downstream gene variant	120	ENST00000558121 <i>el</i> ?	?	1
ICE2 <i>el</i>	downstream gene variant	4207	ENST00000561446 <i>el</i> ?	ENSP00000452713 <i>el</i>	1
ICE2 <i>el</i>	downstream gene variant, upstream gene variant	2229	ENST00000558451 <i>el</i> ?	?	3
ICE2 <i>el</i>	upstream gene variant	3508	ENST00000561144 <i>el</i> ?	?	1
ICE2 <i>el</i>	downstream gene variant	4306	ENST00000561087 <i>el</i> ?	?	1
ICE2 <i>el</i>	downstream gene variant	833	ENST00000560072 <i>el</i> ?	ENSP00000453041 <i>el</i>	3
ICE2 <i>el</i>	downstream gene variant	2897	ENST00000560895 <i>el</i> ?	ENSP00000453820 <i>el</i>	1
ICE2 <i>el</i>	downstream gene variant	1359	ENST00000560406 <i>el</i> ?	ENSP00000453783 <i>el</i>	3
ICE2 <i>el</i>	downstream gene variant	1375	ENST00000560520 <i>el</i> ?	ENSP00000452618 <i>el</i>	3
ICE2 <i>el</i>	downstream gene variant	819	ENST00000558654 <i>el</i> ?	?	3
RORA <i>el</i>	downstream gene variant	222	ENST00000335670 <i>el</i> NM_134261.2	ENSP00000335087 <i>el</i>	3
RORA <i>el</i>	downstream gene variant	4903	ENST00000551975 <i>el</i> ?	ENSP00000449482 <i>el</i>	1
RORA-AS1 <i>el</i>	upstream gene variant	4017	ENST00000559902 <i>el</i> ?	?	1
RP11-745A24.1 <i>el</i>	downstream gene variant	4271	ENST00000475062 <i>el</i> ?	?	1
RP11-745A24.2 <i>el</i>	upstream gene variant	408	ENST00000477729 <i>el</i> ?	?	2
RP11-745A24.3 <i>el</i>	upstream gene variant	1950	ENST00000620477 <i>el</i> ?	?	2

### Putative effect on transcript

#### Intron variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA2 <i>el</i>	ENST00000560936 <i>el</i>	?	?	1
ANXA2 <i>el</i>	ENST00000557906 <i>el</i>	?	ENSP00000452895 <i>el</i>	2
ANXA2 <i>el</i>	ENST00000559113 <i>el</i>	?	ENSP00000453869 <i>el</i>	2
ANXA2 <i>el</i>	ENST00000561445 <i>el</i>	?	?	2
ANXA2 <i>el</i>	ENST00000559467 <i>el</i>	?	ENSP00000453000 <i>el</i>	2
ANXA2 <i>el</i>	ENST00000560466 <i>el</i>	?	ENSP00000453841 <i>el</i>	2
ANXA2 <i>el</i>	ENST00000559559 <i>el</i>	?	?	2
ANXA2 <i>el</i>	ENST00000559370 <i>el</i>	?	ENSP00000453566 <i>el</i>	2
ANXA2 <i>el</i>	ENST00000560367 <i>el</i>	?	ENSP00000453556 <i>el</i>	1
ANXA2 <i>el</i>	ENST00000560468 <i>el</i>	?	ENSP00000452858 <i>el</i>	2

ANXA2 <i>e!</i>	ENST00000421017 <i>e!</i>	NM_001002857.1	ENSP00000411352 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000558132 <i>e!</i>	?	ENSP00000454096 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000332680 <i>e!</i>	NM_001002858.2	ENSP00000346032 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000559956 <i>e!</i>	?	ENSP00000453694 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000558985 <i>e!</i>	?	ENSP00000453039 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000396024 <i>e!</i>	NM_001136015.2	ENSP00000379342 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000557986 <i>e!</i>	?	ENSP00000453742 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000557937 <i>e!</i>	?	?	2
ANXA2 <i>e!</i>	ENST00000559725 <i>e!</i>	?	ENSP00000453174 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000451270 <i>e!</i>	NM_004039.2	ENSP00000387545 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000561022 <i>e!</i>	?	ENSP00000452876 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000558169 <i>e!</i>	?	ENSP00000453945 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000558558 <i>e!</i>	?	ENSP00000452981 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000560165 <i>e!</i>	?	ENSP00000452921 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000558998 <i>e!</i>	?	ENSP00000453925 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000560546 <i>e!</i>	?	?	2
ANXA2 <i>e!</i>	ENST00000559780 <i>e!</i>	?	ENSP00000453770 <i>e!</i>	2
ANXA2 <i>e!</i>	ENST00000557904 <i>e!</i>	?	ENSP00000452958 <i>e!</i>	4
ANXA2 <i>e!</i>	ENST00000559818 <i>e!</i>	?	ENSP00000453859 <i>e!</i>	3
ANXA2 <i>e!</i>	ENST00000560495 <i>e!</i>	?	?	1
ANXA2 <i>e!</i>	ENST00000558503 <i>e!</i>	?	ENSP00000453356 <i>e!</i>	2
ICE2 <i>e!</i>	ENST00000560520 <i>e!</i>	?	ENSP00000452618 <i>e!</i>	1
ICE2 <i>e!</i>	ENST00000558512 <i>e!</i>	?	ENSP00000452714 <i>e!</i>	7
ICE2 <i>e!</i>	ENST00000261520 <i>e!</i>	NM_001018089.2, NM_024611.5	ENSP00000261520 <i>e!</i>	16
ICE2 <i>e!</i>	ENST00000561087 <i>e!</i>	?	?	7
ICE2 <i>e!</i>	ENST00000558121 <i>e!</i>	?	?	8
ICE2 <i>e!</i>	ENST00000560072 <i>e!</i>	?	ENSP00000453041 <i>e!</i>	2
ICE2 <i>e!</i>	ENST00000561124 <i>e!</i>	?	?	8
ICE2 <i>e!</i>	ENST00000561446 <i>e!</i>	?	ENSP00000452713 <i>e!</i>	7
ICE2 <i>e!</i>	ENST00000561328 <i>e!</i>	?	?	3
ICE2 <i>e!</i>	ENST00000560895 <i>e!</i>	?	ENSP00000453820 <i>e!</i>	7
ICE2 <i>e!</i>	ENST00000561114 <i>e!</i>	NM_001276385.1	ENSP00000454162 <i>e!</i>	8
ICE2 <i>e!</i>	ENST00000558181 <i>e!</i>	?	ENSP00000453593 <i>e!</i>	16
ICE2 <i>e!</i>	ENST00000560668 <i>e!</i>	?	ENSP00000453303 <i>e!</i>	7
ICE2 <i>e!</i>	ENST00000560406 <i>e!</i>	?	ENSP00000453783 <i>e!</i>	1
ICE2 <i>e!</i>	ENST00000558654 <i>e!</i>	?	?	2
RORA <i>e!</i>	ENST00000335670 <i>e!</i>	NM_134261.2	ENSP00000335087 <i>e!</i>	1
RORA <i>e!</i>	ENST00000559587 <i>e!</i>	?	?	1
RORA <i>e!</i>	ENST00000449337 <i>e!</i>	NM_134262.2	ENSP00000402971 <i>e!</i>	1
RORA <i>e!</i>	ENST00000309157 <i>e!</i>	NM_002943.3	ENSP00000309753 <i>e!</i>	1
RORA <i>e!</i>	ENST00000261523 <i>e!</i>	NM_134260.2	ENSP00000261523 <i>e!</i>	1
RORA <i>e!</i>	ENST00000501570 <i>e!</i>	?	?	4

RORA-AS1 <i>el</i>	ENST00000501579 <i>el</i>	?	?	4
RORA-AS1 <i>el</i>	ENST00000558140 <i>el</i>	?	?	4
RORA-AS1 <i>el</i>	ENST00000559824 <i>el</i>	?	?	4
RORA-AS1 <i>el</i>	ENST00000558235 <i>el</i>	?	?	4

### 3'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ICE2 <i>el</i>	ENST00000261520 <i>el</i>	NM_001018089.2, NM_024611.5	ENSP00000261520 <i>el</i>	3
ICE2 <i>el</i>	ENST00000561114 <i>el</i>	NM_001276385.1	ENSP00000454162 <i>el</i>	1
ICE2 <i>el</i>	ENST00000558181 <i>el</i>	?	ENSP00000453593 <i>el</i>	3

### 5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA2 <i>el</i>	ENST00000559818 <i>el</i>	?	ENSP00000453859 <i>el</i>	1

### Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ICE2 <i>el</i>	ENST00000558121 <i>el</i>	?	3
ICE2 <i>el</i>	ENST00000561328 <i>el</i>	?	2

