Locus 56 (rs36032232)

SNiPAcard

Block annotations

Block info

genomic range	chr15:60,687,152-60,798,590
block size	111,439 bp
variant count	29 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -0.490 [-3.197 – 1.559]	**gene(s) hit or close-by**	ANXA2, ICE2, RORA, RORA-AS1, RP11-745A24.1, RP11-745A24.2, RP11-745A24.3
phastCons	μ = 0.100 [0 – 0.992]	**eQTL gene(s)**	ICE2
GERP++	μ = -1.141 [-7.15 – 3.05]	**potentially regulated gene(s)**	–
CADD score	μ = 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	ENST00000261520	ILMN_1666610	blood	4.67×10^{-5} (p-value)	Westra et al.	3
ICE2	ENST00000558181					

Putative effect on regulation

Regulatory feature cluster

element id	variant(s)	tissue/cell	factors
ENSR00001452708 (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 (promoter flanking region)	1	cervix (HeLa-S3)	Jund, DNase1
		nervous (NH-A)	DNase1
		A549	H3K4me3
		skin (NHDF-AD)	DNase1
ENSR00000237536	2	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 (enhancer)	1	monocytes (Monocytes-CD14+)	H3K36me3
		A549	H3K4me3, H3K36me3

Variation proximal to gene

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

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

Putative effect on transcript

Intron variant

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

ANXA2	ENST00000421017	NM_001002857.1	ENSP00000411352	2
ANXA2	ENST00000558132	?	ENSP00000454096	2
ANXA2	ENST00000332680	NM_001002858.2	ENSP00000346032	2
ANXA2	ENST00000559956	?	ENSP00000453694	2
ANXA2	ENST00000558985	?	ENSP00000453039	2
ANXA2	ENST00000396024	NM_001136015.2	ENSP00000379342	2
ANXA2	ENST00000557986	?	ENSP00000453742	2
ANXA2	ENST00000557937	?	?	2
ANXA2	ENST00000559725	?	ENSP00000453174	2
ANXA2	ENST00000451270	NM_004039.2	ENSP00000387545	2
ANXA2	ENST00000561022	?	ENSP00000452876	2
ANXA2	ENST00000558169	?	ENSP00000453945	2
ANXA2	ENST00000558558	?	ENSP00000452981	2
ANXA2	ENST00000560165	?	ENSP00000452921	2
ANXA2	ENST00000558998	?	ENSP00000453925	2
ANXA2	ENST00000560546	?	?	2
ANXA2	ENST00000559780	?	ENSP00000453770	2
ANXA2	ENST00000557904	?	ENSP00000452958	4
ANXA2	ENST00000559818	?	ENSP00000453859	3
ANXA2	ENST00000560495	?	?	1
ANXA2	ENST00000558503	?	ENSP00000453356	2
ICE2	ENST00000560520	?	ENSP00000452618	1
ICE2	ENST00000558512	?	ENSP00000452714	7
ICE2	ENST00000261520	NM_001018089.2, NM_024611.5	ENSP00000261520	16
ICE2	ENST00000561087	?	?	7
ICE2	ENST00000558121	?	?	8
ICE2	ENST00000560072	?	ENSP00000453041	2
ICE2	ENST00000561124	?	?	8
ICE2	ENST00000561446	?	ENSP00000452713	7
ICE2	ENST00000561328	?	?	3
ICE2	ENST00000560895	?	ENSP00000453820	7
ICE2	ENST00000561114	NM_001276385.1	ENSP00000454162	8
ICE2	ENST00000558181	?	ENSP00000453593	16
ICE2	ENST00000560668	?	ENSP00000453303	7
ICE2	ENST00000560406	?	ENSP00000453783	1
ICE2	ENST00000558654	?	?	2
RORA	ENST00000335670	NM_134261.2	ENSP00000335087	1
RORA	ENST00000559587	?	?	1
RORA	ENST00000449337	NM_134262.2	ENSP00000402971	1
RORA	ENST00000309157	NM_002943.3	ENSP00000309753	1
RORA	ENST00000261523	NM_134260.2	ENSP00000261523	1
RORA-AS1	ENST00000501570	?	?	4

RORA-AS1	ENST00000501579	?	?	4
RORA-AS1	ENST00000558140	?	?	4
RORA-AS1	ENST00000559824	?	?	4
RORA-AS1	ENST00000558235	?	?	4

3'-UTR variant

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

5'-UTR variant

gene	affected transcript	RefSeq id	protein	variant(s)
ANXA2	ENST00000559818	?	ENSP00000453859	1

Non-coding exon variant

gene	affected transcript	RefSeq id	variant(s)
ICE2	ENST00000558121	?	3
ICE2	ENST00000561328	?	2