

**Table S6.** Summary of genomic annotation for LD surrogates of rs4690351.

FIELD	DESCRIPTION
<b>chr</b>	Chromosome
<b>pos (hg38)</b>	Chromosome position on hg38 obtained from dbSNP build 141
<b>LD (<math>r^2</math>)</b>	Pairwise $r^2$ measure of LD with query SNP from 1000 Genomes Project EUR data
<b>LD (D')</b>	Pairwise D' measure of LD with query SNP from 1000 Genomes Project EUR data
<b>variant</b>	rsID of collected SNP
<b>Ref</b>	Reference nucleotide(s)
<b>Alt</b>	Alternate nucleotide(s)
<b>AFR freq</b>	Allele frequency for African population
<b>AMR freq</b>	Allele frequency for American population
<b>ASN freq</b>	Allele frequency for Asian population
<b>EUR freq</b>	Allele frequency for European population
<b>GERP cons</b>	Conserved regions by GERP (Blue: conserved)
<b>SiPhy cons</b>	Conserved regions by SiPhy (Purple: conserved)
<b>Promoter histone marks</b>	Summary of ChromHMM (core 15-state model) states corresponding to promoter elements from the Roadmap Epigenomics project
<b>Enhancer histone marks</b>	Summary of ChromHMM (core 15-state model) states corresponding to enhancer elements from the Roadmap Epigenomics project
<b>DNase</b>	Summary of DNase hypersensitivity from the Roadmap Epigenomics project
<b>Proteins bound</b>	Protein bound(s) by ChIP-Seq
<b>Motif changed</b>	Motif alteration(s) from TRANSFAC, JASPAR, and protein-binding microarray (PBM) experiments
<b>NHGRI/EBI GWAS hits</b>	SNP in NHGRI GWAS catalog (accessed October 31, 2015)
<b>GRASP QTL hits</b>	SNP in The GRASP Build 2.0.0.0 results and QTLs (non-GWAS)
<b>Selected eQTL hits</b>	SNP in eQTLs obtained from the GTEx analysis V6, the GEUVADIS analysis, and 10 other studies
<b>GENCODE genes</b>	Gene annotations by GENCODE version 13
<b>dbSNP func annot</b>	Functional annotations from dbSNP build 137
<b>RegulomeDB score</b>	RegulomeDB score ( <a href="http://regulomedb.org/help#score">http://regulomedb.org/help#score</a> ). 1 is most likely to be functional, to 7 = least likely.

chr	pos (hg38)	LD (r <sup>2</sup> )	LD (D <sup>2</sup> )	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	GERP cons	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	Motifs changed	NHGRIEBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot	RegulomeDB score	
4	176420424	0.63	0.91	rs4413359	T	A	0.09	0.2	0.28	0.21				SKIN			RFK5			1 hit	896b 3' of SPC33	No Data		
4	176421649	0.63	0.91	rs5076254	T	C	0.15	0.2	0.28	0.21				SKIN			5 altered motifs			1 hit	896b 3' of SPC33	6		
4	176421762	0.63	0.91	rs72710029	G	A	0.14	0.2	0.28	0.21				MUS, SKIN	SKIN		HNFA, Hf			1 hit	906b 3' of SPC33	No Data		
4	176421864	0.63	0.91	rs72710030	T	G	0.09	0.2	0.28	0.21				MUS, SKIN	SKIN				1 hit	906b 3' of SPC33	No Data			
4	176421922	0.63	0.91	rs1114458	C	T	0.15	0.2	0.28	0.21				MUS, SKIN	SKIN		12 altered motifs			1 hit	906b 3' of SPC33	6		
4	176423068	0.63	0.91	rs5864374	CT	C	0.12	0.2	0.28	0.21				ESDR, BRN, SKIN			6 altered motifs			1 hit	916b 3' of SPC33	No Data		
4	176423664	0.63	0.91	rs56114616	C	T	0.14	0.2	0.27	0.21				ESDR, SKIN			7 altered motifs			1 hit	916b 3' of SPC33	5		
4	176424548	0.63	0.91	rs897042	G	A	0.16	0.2	0.28	0.21				MUS, SKIN						1 hit	926b 3' of SPC33	6		
4	176425362	0.63	0.91	rs5742321	C	T	0.14	0.2	0.28	0.21				MUS, SKIN, BONE						1 hit	936b 3' of SPC33	4		
4	176425925	0.61	0.9	rs3969905	T	A	0.14	0.2	0.28	0.21							9 altered motifs			1 hit	936b 3' of SPC33	5		
4	176426609	0.63	0.91	rs72710032	C	T	0.07	0.19	0.27	0.21				ESDR, IPSC, BRN						1 hit	946b 3' of SPC33	5		
4	176426616	0.63	0.91	rs72710033	G	T	0.14	0.2	0.27	0.21				ESDR, IPSC, BRN			5 altered motifs			1 hit	946b 3' of SPC33	5		
4	176427316	0.64	0.92	rs72710034	G	A	0.13	0.2	0.25	0.21							9 altered motifs			1 hit	956b 3' of SPC33	6		
4	176427842	0.64	0.92	rs76895402	T	C	0.15	0.2	0.25	0.21							Pax-4			1 hit	966b 3' of SPC33	No Data		
4	176428246	0.64	0.92	rs17062905	T	C	0.15	0.2	0.25	0.21				BRN, BRN			NF-Y, Sox			1 hit	966b 3' of SPC33	5		
4	176429167	0.64	0.92	rs2122334	G	T	0.13	0.2	0.25	0.21							5 altered motifs			1 hit	976b 3' of SPC33	No Data		
4	176429327	0.64	0.92	rs2122335	T	C	0.15	0.2	0.25	0.21							8 altered motifs			1 hit	976b 3' of SPC33	No Data		
4	176429542	0.64	0.92	rs2167094	T	C	0.15	0.2	0.25	0.21							8 altered motifs			1 hit	976b 3' of SPC33	5		
4	176429839	0.64	0.92	rs4690438	T	C	0.13	0.2	0.25	0.21							Pou2f2, Pou6f1			1 hit	986b 3' of SPC33	No Data		
4	176429967	0.64	0.92	rs4690348	A	G	0.15	0.2	0.25	0.21							HIF1, UFH3BETA			1 hit	986b 3' of SPC33	No Data		
4	176430061	0.64	0.92	rs1377835	G	C	0.09	0.2	0.25	0.21							10 altered motifs				986b 3' of SPC33	6		
4	176430171	0.63	0.91	rs1377836	T	C	0.13	0.2	0.25	0.21							GR, NRSF			1 hit	986b 3' of SPC33	No Data		
4	176430618	0.62	0.91	rs72710037	C	T	0.15	0.2	0.25	0.21										1 hit	986b 3' of SPC33	No Data		
4	176431093	0.64	0.92	rs1869901	T	C	0.15	0.2	0.25	0.21							Ets, Pax-2			1 hit	986b 3' of SPC33	No Data		
4	176431402	0.64	0.92	rs1869902	T	C	0.15	0.2	0.25	0.21							Foxa, SETDB1, YY1			1 hit	986b 3' of SPC33	No Data		
4	176431762	0.64	0.92	rs2877951	T	A	0.13	0.2	0.25	0.21							7 altered motifs			1 hit	1006b 3' of SPC33	6		
4	176431945	0.63	0.92	rs55804250	G	A	0.15	0.2	0.25	0.21							GZF1, TATA, TGF4			1 hit	1006b 3' of SPC33	No Data		
4	176431953	0.64	0.92	rs60121301	C	A	0.13	0.2	0.25	0.21							BCI, Hf			1 hit	1006b 3' of SPC33	No Data		
4	176432165	0.64	0.92	rs17062918	T	C	0.15	0.2	0.25	0.21							Me2, Pou2f2, TATA			1 hit	1006b 3' of SPC33	No Data		
4	176432672	0.64	0.92	rs6811555	C	T	0.15	0.2	0.25	0.21				BRN						1 hit	1006b 3' of SPC33	6		
4	176432995	0.64	0.92	rs17062929	A	G	0.13	0.2	0.25	0.21				STRM, BRN	HRT, BRN, LING		Foxp3, Pou5f1			1 hit	1016b 3' of SPC33	5		
4	176433136	0.64	0.92	rs4690439	A	G	0.15	0.2	0.25	0.21				STRM, BRN	LING		BDP1, En-1, GR			1 hit	1016b 3' of SPC33	5		
4	176433200	0.64	0.92	rs4690440	G	C	0.09	0.2	0.25	0.21				STRM, BRN	LING						1 hit	1016b 3' of SPC33	5	
4	176433689	0.64	0.92	rs11732434	T	C	0.15	0.2	0.25	0.21				4 tissues			5 altered motifs			1 hit	1016b 3' of SPC33	6		
4	176437461	0.67	0.93	rs4690442	G	A	0.06	0.19	0.17	0.21				5 tissues								1056b 3' of SPC33	6	
4	176439518	0.66	-0.93	rs921691	G	A	0.79	0.81	0.83	0.79				ESDR, SKIN, CRVX			Evi-1, Foxa, Hand1		1 hit		1076b 3' of SPC33	No Data		
4	176440515	0.66	-0.93	rs7654706	T	A	0.78	0.8	0.83	0.79							6 altered motifs				1086b 3' of SPC33	5		
4	176442611	0.66	-0.93	rs6825079	T	G	0.78	0.8	0.83	0.79				9 tissues	LING, MUS							1106b 3' of SPC33	5	
4	176443053	0.66	-0.93	rs6826013	T	G	0.79	0.8	0.83	0.79				9 tissues	SKIN, MUS		4 altered motifs					1116b 3' of SPC33	3a	
4	176443250	0.67	-0.93	rs309755	C	G	0.9	0.81	0.83	0.79				9 tissues	SKIN, LING	5 bound proteins	ILF2, FOXA, BAF155, SRSX, Znf14					1116b 3' of SPC33	4	
4	176443365	0.66	-0.93	rs5553962	T	C	0.79	0.8	0.83	0.79				9 tissues	SKIN, LING		6 altered motifs					1136b 3' of SPC33	6	
4	176445360	0.67	0.93	rs17062956	A	G	0.06	0.19	0.17	0.21				9 tissues			10 altered motifs					1146b 3' of SPC33	6	
4	176446121	0.65	-0.93	rs5864377	TG	T	0.78	0.8	0.82	0.79							PAC1, NR4B1-1, TATA					1196b 3' of SPC33	No Data	
4	176451356	0.66	-0.94	rs1016568	A	G	0.78	0.72	0.8	0.79							Hsf, STAT					1216b 3' of SPC33	4	
4	176453492	0.68	-0.94	rs309753	C	A	0.89	0.72	0.8	0.79				ESDR, BRN			Hf, Pou3f3					1276b 3' of SPC33	6	
4	176458924	0.69	0.94	rs4234857	T	C	0.06	0.28	0.2	0.21							Pou5f1					1296b 3' of SPC33	6	
4	176461191	0.68	-0.94	rs309708	T	C	0.89	0.73	0.8	0.79							Foxp3, TCF12, ZEB1					1306b 3' of SPC33	No Data	
4	176461971	0.68	-0.95	rs5553966	T	C	0.77	0.72	0.8	0.79							Pou5f1					1306b 3' of SPC33	5	
4	176462123	0.69	-0.95	rs309709	A	G	0.86	0.72	0.8	0.79							9 altered motifs					1306b 3' of SPC33	No Data	
4	176464240	0.79	0.94	rs3688277	C	G	0.17	0.25	0.2	0.18				BLD		BCI11A, PU1	Eomes, Foxo1, Sox					1326b 3' of SPC33	6	
4	176465049	0.79	0.94	rs6553967	C	A	0.1	0.25	0.19	0.18				BLD	BLD		GATA, NF-1					1336b 3' of SPC33	2b	
4	176467622	0.8	0.94	rs17063033	T	C	0.07	0.25	0.19	0.18				PLCNT			Evi-1, Pax3, Znf143					1356b 3' of SPC33	No Data	
4	176468553	0.64	-0.94	rs10034533	C	T	0.73	0.72	0.75	0.78												1366b 3' of SPC33	No Data	
4	176468942	0.8	-0.95	rs432164	T	A	0.79	0.75	0.81	0.82				STRM			GATA					1386b 3' of SPC33	No Data	
4	176470069	0.8	-0.95	rs364538	T	C	0.8	0.75	0.81	0.82							CTCF, Evi-1, Rad21					1386b 3' of SPC33	6	
4	176470305	0.81	0.95	rs12796082	C	T	0.13	0.26	0.2	0.18							p300					1386b 3' of SPC33	No Data	
4	176471509	0.8	-0.95	rs423719	G	A	0.82	0.75	0.81	0.82							STAT					1386b 3' of SPC33	No Data	
4	176473089	0.81	0.95	rs72710075	T	C	0.07	0.25	0.19	0.18							10 altered motifs					1416b 3' of SPC33	5	
4	176478200	0.96	1	rs72710080	T	C	0.1	0.17	0.12	0.16				5 tissues	ESDR, SKIN, MUS		GR					1466b 3' of SPC33	4	
4	176480851	1	1	rs11731341	C	T	0.13	0.16	0.13	0.17							4 altered motifs					1506b 3' of SPC33	No Data	
4	176482720	1	1	rs35730736	AT	A	0.13	0.16	0.13	0.17							25 altered motifs					1506b 3' of SPC33	6	
4	176483591	1	1	rs17063122	A	T	0.08	0.16	0.13	0.17							4 altered motifs					1516b 3' of SPC33	6	
4	176485562	0.58	1	rs7545370	10-mer	G	0.14	0.16	0.13	0.16							4 altered motifs					1536b 3' of SPC33	No Data	
4	176489462	1	1	rs11133155	G	A	0.08	0.16	0.13	0.17				SKIN								1576b 3' of SPC33	No Data	
4	176489857	1	1	rs11133156	G	A	0.06	0.16	0.13	0.17							Hoxa5					1586b 3' of SPC33	No Data	
4	176489859	1	1	rs11133157	G	C	0.11	0.16	0.13	0.17							Hoxa5					1586b 3' of SPC33	No Data	
4	176490693	1	1	rs72710094	T	G	0.1	0.16	0.13	0.17							Foxo1, Hf, ZEB1					1586b 3' of SPC33	6	
4	176493741	1	1	rs72710095	A																			