

Supplementary Table S3. Functional annotations of SNPs in high LD ($r^2 > 0.80$) with rs3791686 based on HaploReg V 4.1 and RegulomeDB Score

Chr	Position (hg1)	LD (r^2) ^a	LD (D) ^a	Variant	Ref	Alt	AFR Freq	AMR Freq	ASN Freq	EUR Freq	HaploReg V 4.1										RegulomeDB	
											SiPhy Conserved	Promoter Histone	Enhancer Histone	DNase	Proteins Bound	Motifs Changed	NHGRI/EBI GWAS Hits	GRASP QTL Hits	Selected eQTL Hits	GENCODE Genes	dbSNP functional	RegulomeDB Score ^b
2	212221868	0.81	0.91	rs2033644	T	G	0.09	0.31	0.28	0.27	No								19kb 3' of ERBB4		6	
2	212221870	0.81	0.91	rs2033643	C	A	0.09	0.31	0.28	0.27	No								19kb 3' of ERBB4		6	
2	212236888	0.87	0.95	rs12992319	G	A	0.17	0.3	0.27	0.28	No								Foxl1, ZEB1		7	
2	212238982	0.9	0.95	rs1821659	T	C	0.22	0.31	0.28	0.27	No								Bel6b, STAT		7	
2	212239070	0.88	0.94	rs1821658	C	G	0.22	0.31	0.28	0.28	No								7 altered motifs		6	
2	212242434	0.89	0.96	rs7565960	G	A	0.16	0.28	0.28	0.26	No								21 altered motifs		6	
2	212243703	0.89	0.96	rs13003941	G	T	0.11	0.31	0.28	0.32	No								6 altered motifs		4	
2	212251864	0.89	0.96	rs3748962	T	C	0.08	0.31	0.29	0.32	Yes								PANC		7	
2	212252955	0.92	0.97	rs934606	C	T	0.08	0.31	0.28	0.31	No								ESC		7	
2	212258353	1	1	rs3791686	T	C	0.29	0.33	0.28	0.34	No								ESDR	ESDR		6
2	212259174	1	1	rs1836720	C	G	0.34	0.35	0.28	0.34	No								ESDR, BRN		5	
2	212259228	1	1	rs1836719	A	G	0.34	0.32	0.28	0.32	No								ESDR, PANC		5	
2	212267719	0.84	0.97	rs12998535	C	T	0.08	0.28	0.26	0.27	No								GR		7	
2	212274937	0.88	0.95	rs3828242	A	G	0.07	0.28	0.28	0.28	No								4 tissues	BRN		6
																			Hoxa5, SIX5, Sox		6	

^aLD was calculated from the 1000 Genome Projects (East Asian).

^bRegulomeDB Score was categorized as follows: 4, TF binding + DNase peak; 5, TF binding or DNase peak; 6, Motif hit; 7, no data (Alan et al. 2012).

Abbreviations: Chr, chromosome; LD, linkage disequilibrium; Ref, reference allele; Alt, alternate non-reference allele; Freq, frequency; AFR, African; AMR, Ad Mixed American; ASN, East Asian, EUR, European; PANC, pancreatic Islets; ESC, embryonic stem (ES) cells; ESDR, ES-derived cells; BRN, brain tissues; DNase, deoxyribonuclease; QTL, quantitative trait loci; eQTL, expression quantitative trait loci; TF, transcription factor.