

Figure S1. Full screen shot from HaploReg (<http://www.broadinstitute.org/mammals/haploreg/haploreg.php>, last accessed on 03/20/2014), when three xMHC SNPs (IDs in red) being highlighted in this study are queried for functional relevance and LD patterns in populations of African ancestry.

HaploReg v2

HaploReg is a tool for exploring annotations of the noncoding genome at variants on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci. Using LD information from the 1000 Genomes Project, linked SNPs and small indels can be visualized along with their predicted chromatin state, their sequence conservation across mammals, and their effect on regulatory motifs. HaploReg is designed for researchers developing mechanistic hypotheses of the impact of non-coding variants on clinical phenotypes and normal variation.

Update 2013.02.14: Version 2 now includes an expanded library of SNPs (based on dbSNP 137), motif instances (based on PWMs discovered from ENCODE experiments), enhancer annotations (adding 90 cell types from the Roadmap Epigenome Mapping Consortium), and eQTLs (from the GTex eQTL browser). In addition, LD calculations are provided based on the 1000 Genomes Phase 1 individuals, and r^2 and D' measurements are available down to an r^2 threshold of 0.2. Display improvements include improved cell metadata, gene metadata, and PWM display on the detail pages and the option for text output. Version 1 is available [here](#).

Build Query **Set Options** **Documentation**

Use one of the three methods below to enter a set of variants. If an r^2 threshold is specified (see the Set Options tab), results for each variant will be shown in a separate table along with other variants in LD. If r^2 is set to NA, only queried variants will be shown, together in one table.

Query (comma-delimited list of rsIDs OR a single region as chrN:start-end):

or, upload a text file (one refSNP ID per line):

or, select a GWAS:

Enhancer enrichment analysis
No significant enrichment

DNase enrichment analysis
No significant enrichment

Query SNP: **rs3094626** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	30323623	1	1	rs3094626	T	C	0.38	0.36	0.45	0.37							Nrf-2,PRDM1	9kb 3' of RPP21	

Query SNP: **rs3134931** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	32190620	1	1	rs3134931	T	C	0.45	0.36	0.56	0.31			HMEC, Huvec, GM12878	Th1,Fibrobl,Osteobl	PU1		p300	NOTCH4	intronic

Query SNP: **rs2857114** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	32779996	1	1	rs2857114	G	A	0.51	0.59	0.72	0.66							9 altered motifs	543bp 3' of HLA-DOB	
6	32780470	1	1	rs2621331	C	T	0.51	0.58	0.67	0.66							8 altered motifs	69bp 3' of HLA-DOB	

Figure S2. Partial screen shot from HaploReg (<http://www.broadinstitute.org/mammals/haploreg/haploreg.php>, last accessed on 03/20/2014), when three xMHC SNPs (IDs in red) being highlighted in this study are queried for functional relevance and LD patterns in populations of European ancestry.

Query SNP: **rs3094626** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	30323623	1	1	rs3094626	T	C	0.38	0.36	0.45	0.37							Nrf-2,PRDM1	9kb 3' of RPP21	
6	30338247	0.8	0.95	rs3129813	G	A	0.44	0.35	0.44	0.34							Hand1,Ncx,RFX5	24kb 3' of RPP21	
6	30338248	0.8	0.95	rs3130357	A	G	0.44	0.35	0.44	0.34							Hand1	24kb 3' of RPP21	

Query SNP: **rs3134931** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	32190620	1	1	rs3134931	T	C	0.45	0.36	0.56	0.31			HMEC, Huvec, GM12878	Th1,Fibrobl,Osteobl	PU1		p300	NOTCH4	intronic

Query SNP: **rs2857114** and variants with $r^2 \geq 0.8$

chr	pos (hg19)	LD (r ²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNase	Proteins bound	eQTL tissues	Motifs changed	GENCODE genes	dbSNP func annot
6	32740761	0.81	0.93	rs2621420	T	C	0.65	0.64	0.74	0.68			HMEC, NHEK				HDAC2,SP1B	9.4kb 5' of HLA-DQB2	
6	32760249	0.92	0.96	rs2857160	T	G	0.64	0.61	0.73	0.66							8 altered motifs	20kb 3' of HLA-DOB	
6	32760257	0.94	0.98	rs2857159	T	C	0.64	0.61	0.73	0.67							4 altered motifs	20kb 3' of HLA-DOB	
6	32760334	0.95	0.99	rs2857158	A	G	0.64	0.61	0.73	0.67							5 altered motifs	20kb 3' of HLA-DOB	
6	32760539	0.95	0.99	rs2187687	T	C	0.64	0.61	0.73	0.67							11 altered motifs	20kb 3' of HLA-DOB	
6	32760965	0.95	0.99	rs2894294	G	A	0.64	0.61	0.72	0.67			H7-hESC,NT2-D1				Egr-1	20kb 3' of HLA-DOB	
6	32761317	0.96	1	rs2857155	A	G	0.64	0.62	0.72	0.67							14 altered motifs	19kb 3' of HLA-DOB	
6	32761507	0.96	1	rs2187686	A	G	0.62	0.62	0.73	0.67							Hoxb8	19kb 3' of HLA-DOB	
6	32761776	0.96	1	rs2157081	G	A	0.64	0.62	0.72	0.67							4 altered motifs	19kb 3' of HLA-DOB	
6	32761826	0.96	1	rs2157080	T	C	0.64	0.62	0.72	0.67							10 altered motifs	19kb 3' of HLA-DOB	
6	32762103	0.96	1	rs2157079	T	C	0.64	0.62	0.72	0.67			GM12878					18kb 3' of HLA-DOB	
6	32762226	0.95	0.98	rs2157078	A	G	0.64	0.61	0.72	0.67			GM12878				HDAC2,p300	18kb 3' of HLA-DOB	
6	32762307	0.96	1	rs2067098	T	C	0.64	0.62	0.72	0.67			GM12878	Adult_CD4_Th0				18kb 3' of HLA-DOB	
6	32762484	0.96	1	rs2067577	C	G	0.64	0.62	0.72	0.67			GM12878	7 cell types	PU1		Foxo	18kb 3' of HLA-DOB	
6	32762616	0.96	1	rs2857154	A	G	0.64	0.62	0.72	0.67			GM12878	CD20+			Myc	18kb 3' of HLA-DOB	
6	32762654	0.96	1	rs2857152	A	G	0.64	0.62	0.72	0.67			GM12878	CD20+			Foxc1,Hbp1,Nlx3	18kb 3' of HLA-DOB	
6	32763040	0.96	1	rs2621378	G	A	0.64	0.62	0.72	0.67							4 altered motifs	17kb 3' of HLA-DOB	
6	32763514	0.96	1	rs2857151	A	G	0.64	0.62	0.72	0.67					MAFK		En-1,Pax-5	17kb 3' of HLA-DOB	
6	32763521	0.96	1	rs2621376	T	C	0.64	0.62	0.72	0.67					MAFK		En-1,RFX5	17kb 3' of HLA-DOB	
6	32763821	0.96	1	rs2857150	A	G	0.64	0.62	0.72	0.67							29 altered motifs	17kb 3' of HLA-DOB	
6	32763974	0.95	0.98	rs2621375	T	C	0.64	0.61	0.72	0.67							Zbtb3	17kb 3' of HLA-DOB	
6	32764719	0.96	1	rs2187684	C	T	0.63	0.62	0.72	0.67			8 cell types	H1			Mef2,Pou2f2	16kb 3' of HLA-DOB	
6	32765383	0.96	1	rs2257789	C	A	0.63	0.62	0.72	0.67			8 cell types	H1, NHEK	28 cell types	PU1,ELF1	Spz1	15kb 3' of HLA-DOB	
6	32767091	0.96	1	rs2621371	G	T	0.63	0.62	0.72	0.67			GM12878	Osteobl		PU1,POL2	8 altered motifs	13kb 3' of HLA-DOB	
6	32770390	0.96	1	rs1600571	G	A	0.63	0.62	0.72	0.67							ATF2,E4BP4,Hsf	10kb 3' of HLA-DOB	
6	32771354	0.96	1	rs2621354	T	G	0.63	0.62	0.72	0.67					PU1		Bcl6b	9.2kb 3' of HLA-DOB	
6	32771531	0.92	0.96	rs2621353	G	C	0.63	0.60	0.72	0.66								9kb 3' of HLA-DOB	
6	32771610	0.96	1	rs2857144	A	G	0.63	0.62	0.72	0.67							Hmbox1	8.9kb 3' of HLA-DOB	
6	32771887	0.96	1	rs2621352	C	T	0.63	0.62	0.72	0.67							AP-2,NF-1,PTF1-beta	8.7kb 3' of HLA-DOB	
6	32772004	0.96	1	rs2621351	G	T	0.63	0.62	0.72	0.67							p300	8.5kb 3' of HLA-DOB	
6	32772086	0.86	0.95	rs202007295	TTG	T	0.59	0.60	0.68	0.66							10 altered motifs	8.5kb 3' of HLA-DOB	
6	32772087	0.93	0.98	rs138384239	TG	T	0.61	0.62	0.71	0.67							17 altered motifs	8.5kb 3' of HLA-DOB	
6	32772504	0.91	0.96	rs200016005	G	GCT	0.61	0.62	0.72	0.67								8kb 3' of HLA-DOB	
6	32772505	0.91	0.96	rs200139770	C	CTCT	0.61	0.63	0.72	0.67							CTCF	8kb 3' of HLA-DOB	
6	32772556	0.94	0.98	rs2621350	C	A	0.62	0.61	0.71	0.67								8kb 3' of HLA-DOB	
6	32772564	0.84	0.97	rs2621349	G	C	0.58	0.59	0.69	0.64							TCF4	8kb 3' of HLA-DOB	
6	32772575	0.84	0.96	rs2621348	T	G	0.59	0.59	0.71	0.64							Foxp1,RREB-1	8kb 3' of HLA-DOB	