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Supplemental Data

Two Functional Lupus-Associated *BLK* Promoter Variants Control Cell-Type- and Developmental-Stage-Specific Transcription

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Figure S1.

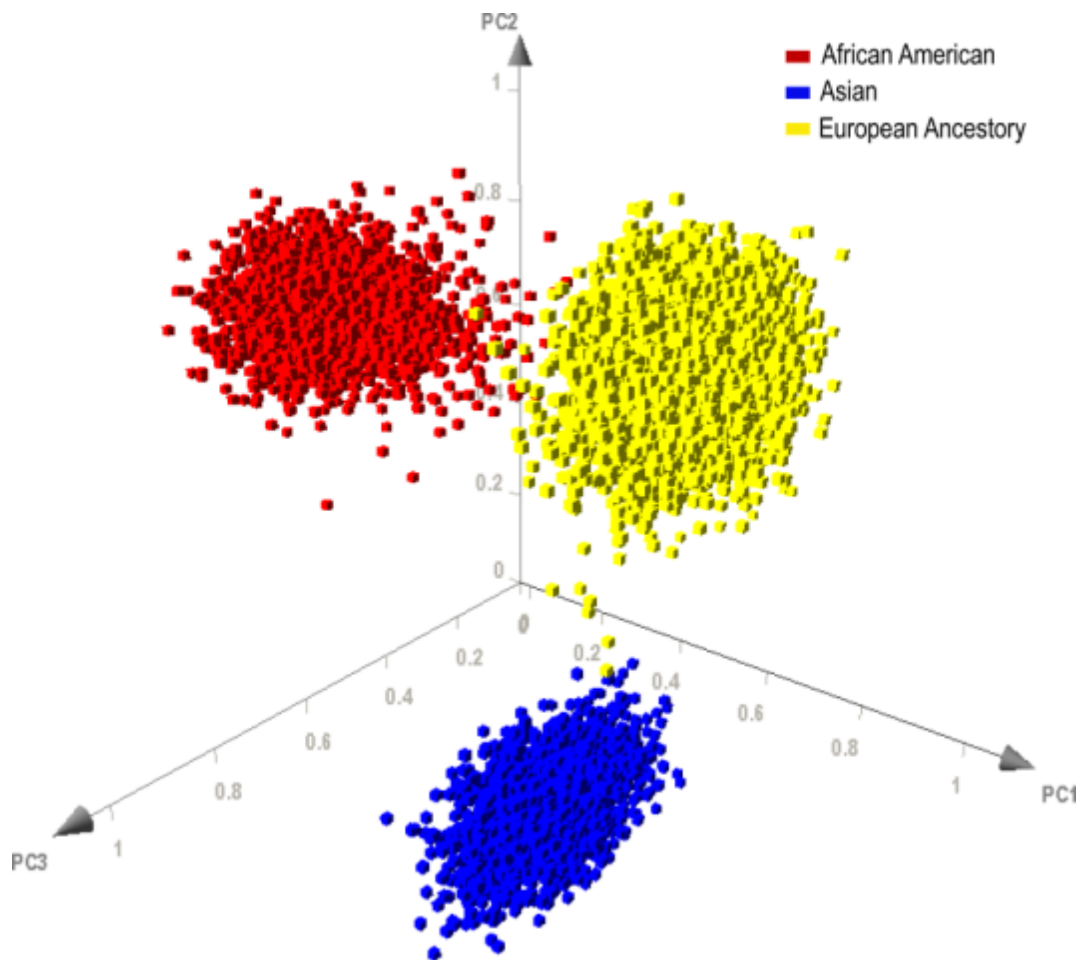


Figure S1. Population substructure of study participants used in genetic association analysis.

The population segregation based on the first three principle components calculated for this dataset are shown. Red = African-American; Blue = Asian; Yellow = European-American

Figure S2.

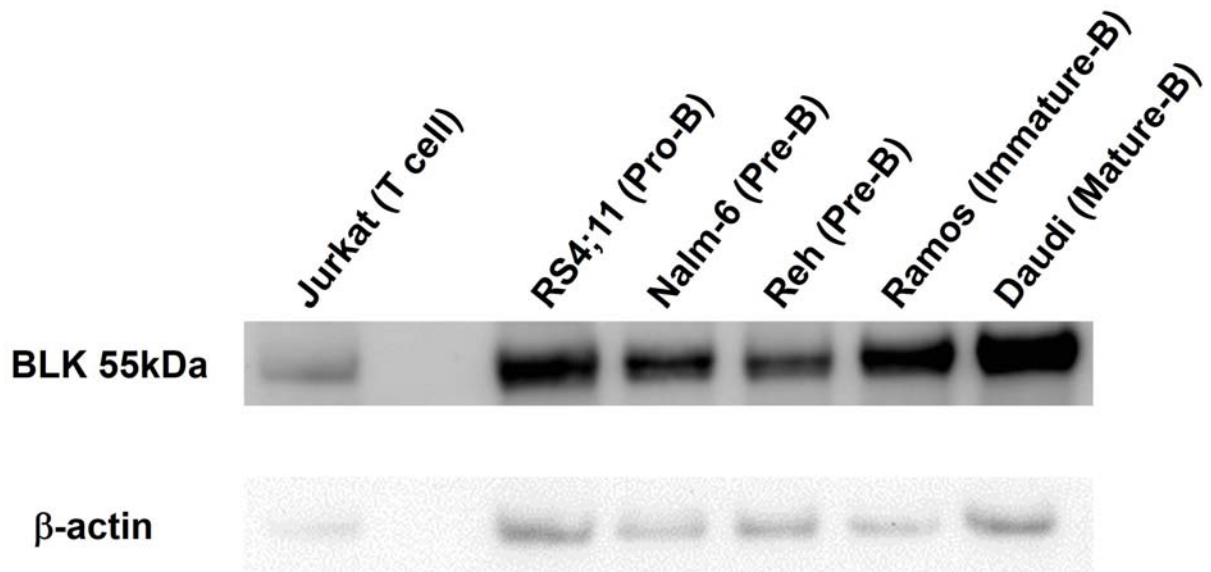


Figure S2: BLK protein expression in the cell lines used in reporter assays. The 55 kDa BLK band was detected in all cell lines used in the study using the anti-human BLK antibody (clone 7A12, Novus Biologicals). The antibody has been shown to specifically bind to BLK native and recombinant proteins.

Figure S3.

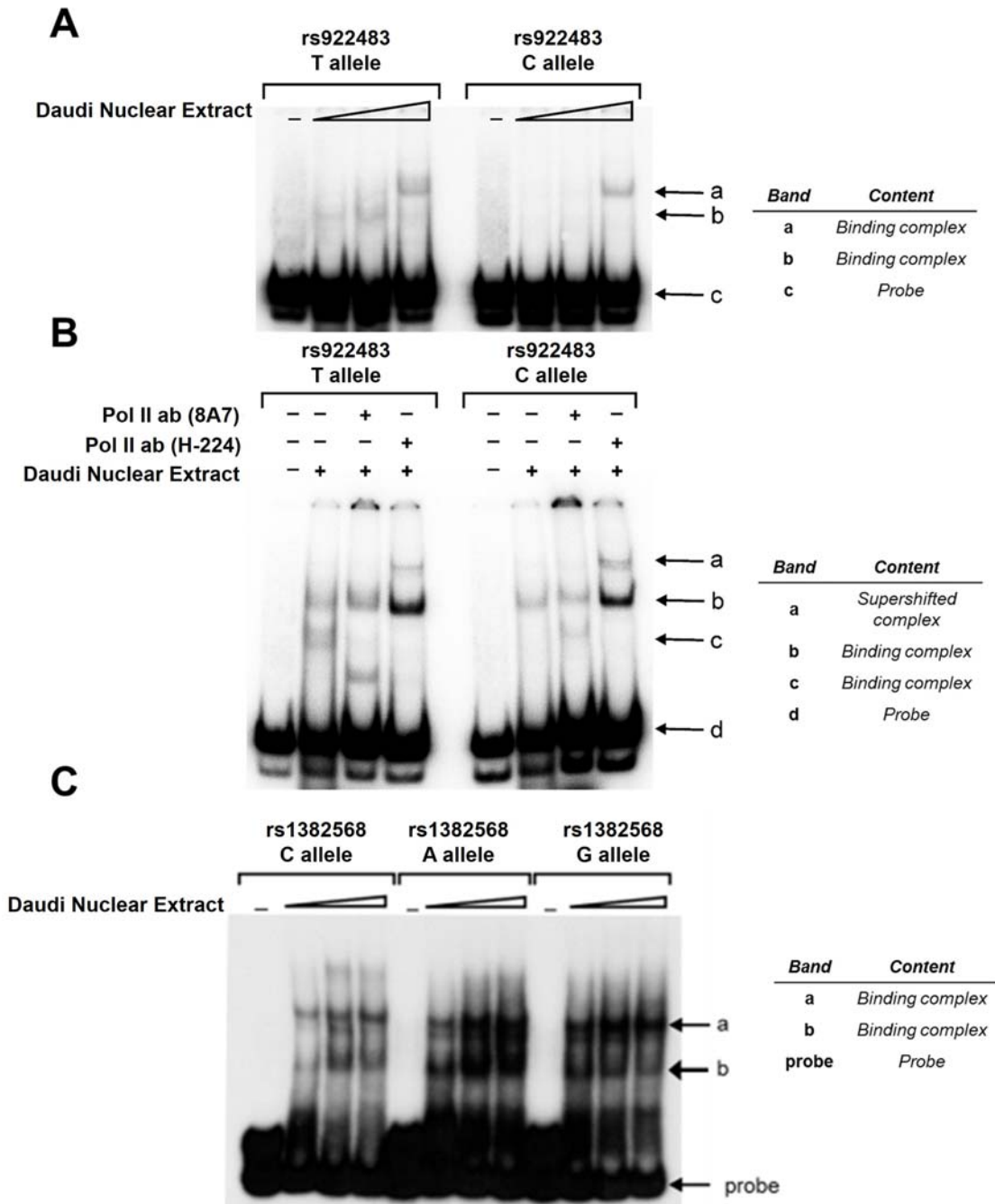


Figure S3: Risk alleles at rs922483 and rs1382568 influence binding of specific transcriptional factors (TFs) complexes. Electrophoretic mobility shift assay confirming differences in nuclear

factor binding of Probe P1 (100 bp) flanking the P1 SNP variant (rs922483) is shown in panel (A). “-” represents labeled DNA probe only control. Subsequent lanes represent the binding of increasing amounts of nuclear factors extracted from Daudi nuclei. Supershift assay of P1 probe is shown in panel (B). Two ug of anti-RNAPII antibodies were co-incubated with nuclear factor prior to the addition of labeled probes. The electrophoretic mobility shift of probe P2 flanking P2 SNP (rs1382568) is shown in panel (C). Increasing amount of nuclear factors extracted from Daudi nuclei were used to titrate the binding affinity. All results shown are of a representative experiment.

Figure S4.

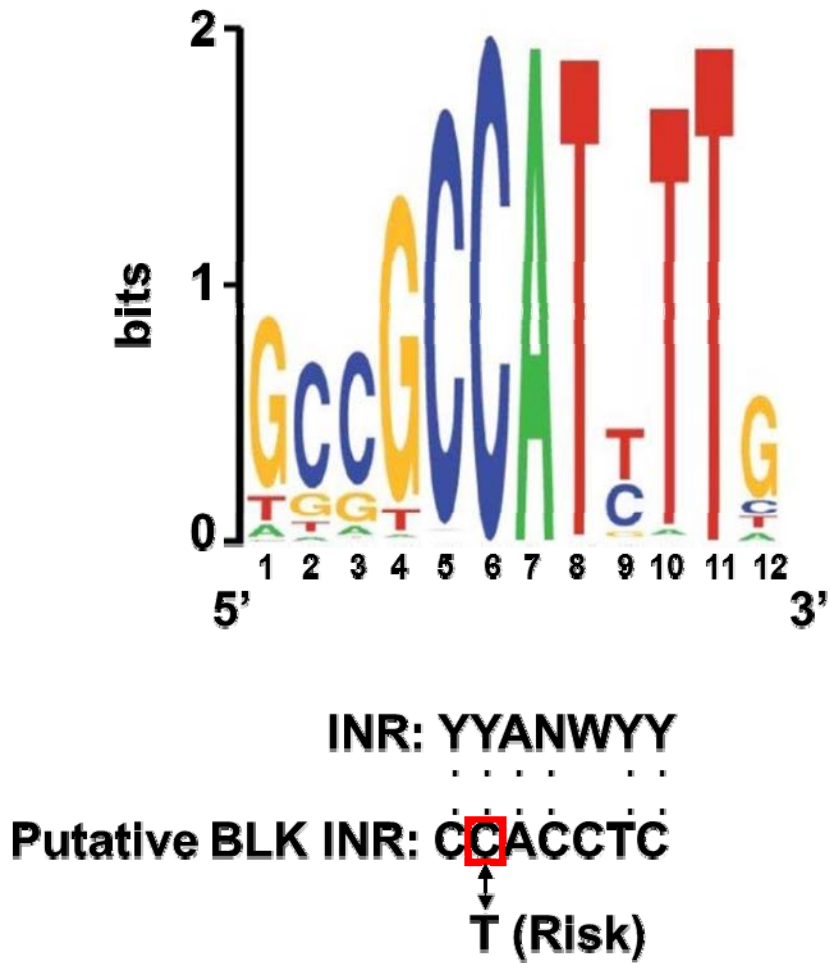


Figure S4. The variant rs922483 resides within a human Initiation (INR) motif. A weblogo diagram (<http://weblogo.berkeley.edu/logo.cgi>) based on alignments of INR motifs done by Yarden et.al.¹ and the corresponding *BLK* putative INR sequence with the location of variant rs922483 is shown (red box). *BLK* sequence location is shown in **Figure 1D**.

Table S1: Source and demographics of samples used in the SNP genotyping association analyses presented.

Contributor/Group	<i>European-Ancestry</i>		<i>Asian</i>		<i>African-American</i>		<i>Total</i>	
	Case	Control	Case	Control	Case	Control	Case	Control
Stevens: United States (Washington)	19	0	10	0	2	0	31	0
Tsao: United States (California), China	30	0	534	483	70	6	634	489
Jacob: United States (California)	108	0	82	0	0	0	190	0
^a PROFILE: Brown: United States (Alabama)	731	1083	0	0	575	637	1306	1720
CLU & SLEIGH: Gilkeson, Kamen: United States (South Carolina)	73	175	0	0	112	65	185	240
^b LFRR/ORDRCC: Harley, James: United States (Oklahoma)	667	553	0	0	428	209	1095	762
^c LuMNS: Gaffney, Sivils : United States (Minnesota)	445	0	0	0	0	0	445	0
ORDRCC: James: United States (Oklahoma)	122	137	2	46	52	55	176	238
Merrill: United States (Oklahoma)	78	2	0	0	28	5	106	7
Vyse: England	248	528	0	0	0	0	248	528
Scofield: United States (Oklahoma)	30	66	0	0	0	0	30	66
^d TRIDOM: Niewold: United States (Illinois, Minnesota)	79	0	0	0	139	0	218	0
Criswell: United States (California)	79	0	0	0	0	0	79	0
Boackle: United States (Colorado)	14	0	2	0	0	0	16	0
^e Gregersen: United States (New York)	0	0	0	0	0	299	0	299
Freedman: United States (North Carolina)	0	0	0	0	0	458	0	458
Bae: South Korea	0	0	642	741	0	0	642	741
BIOLUPUS: Sweden	78	0	0	0	0	0	78	0
BIOLUPUS: Belgium	39	53	0	0	0	0	39	53
Pons-Estel: Argentina (Europeans)	44	0	0	0	0	0	44	0
BIOLUPUS: Spain	464	419	0	0	0	0	464	419
BIOLUPUS: Portugal	186	174	0	0	0	0	186	174
BIOLUPUS: Hungary	25	47	0	0	0	0	25	47
BIOLUPUS: Italy	339	309	0	0	0	0	339	309
BIOLUPUS: Holland	65	0	0	0	0	0	65	0
BIOLUPUS: Danish	17	0	0	0	0	0	17	0
Totals:	3980	3546	1272	1270	1406	1734		

^aPROFILE (US multi-ethnic, multi-regional cohort of lupus patients), ^bLFRR (Lupus Family Registry and Repository), ^cLuMNAS (Univ. of Minnesota Lupus Association Cohort), ^dTRIDOM (University of Chicago Translational Research in the Department of Medicine registry), ^eFeinstein Institute of Medical Research (laboratory of Dr. Gregersen).

Table S2: Demographics of the ABCoN/NYCP re-sequencing samples

Ancestry	Case			Control		
	<i>Male</i>	<i>Female</i>	Total	<i>Male</i>	<i>Female</i>	Total
European-Ancestry	0	191	191	0	96	96

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Supplemental References

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