Supplementary Table 1. Association of rs73366469 with SLE detected by the Immunochip

Immunochip SNP at the *GTF2IRD1-GTF2I* region	Position on Chr7 (hg19)	Allele	Chinese (1010 cases and 848 controls)						European American (930 and 1107)					African American (532 and 367)				
			MAF (%)					r^2 with	MAF (%)					MAF (%)				
			SLE	Control	*P*	OR	95%CI	rs73366469	SLE	Control	*P*	OR	95%CI	SLE	Control	*P*	OR	95%CI
rs10252087	73925395	G/A	3.7	4.4	0.19	0.80	0.57-1.12	0.00	--	--	--	--	--	--	--	--	--	--
rs9638190	73934634	G/A	36.4	39.1	0.03	0.86	0.75-0.98	0.02	--	--	--	--	--	--	--	--	--	--
rs10261551	73945039	0/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs59656369	73949411	C/T	36.5	33.9	0.22	1.09	0.95-1.26	0.00	--	--	--	--	--	--	--	--	--	--
rs757575	73961074	T/C	4.7	5.9	0.12	0.79	0.59-1.06	0.00	--	--	--	--	--	--	--	--	--	--
rs73137125	74018950	G/A	6.9	8.5	0.02	0.75	0.58-0.96	0.02	--	--	--	--	--	--	--	--	--	--
imm-7-73657728	74019792	0/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs10262535	74021499	T/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs13244257	74024788	0/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs112970834	74024915	T/C	0.0	0.1	0.94	0.88	0.04-18.33	0.01	--	--	--	--	--	--	--	--	--	--
rs76239959	74029672	C/A	0.1	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs6460086	74029742	G/T	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs73702631	74030842	T/C	4.8	6.0	0.10	0.78	0.58-1.05	0.02	--	--	--	--	--	--	--	--	--	--
rs77202747	74031787	T/A	0.0	0.1	0.58	0.49	0.04-6.36	0.01	--	--	--	--	--	--	--	--	--	--
rs118073108	74032575	C/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs73366469	74033600	C/T	28.7	12.6	3.8E-29	2.88	2.39-3.47	1.00	12.5	9.4	7.5E-03	1.32	1.08-1.61	17.7	19.6	0.32	0.88	0.69-1.13
rs75170648	74033723	C/A	0.7	2.3	2.7E-03	0.37	0.20-0.71	0.00	--	--	--	--	--	--	--	--	--	--
rs77922881	74034068	A/G	0.0	0.1	0.66	0.53	0.03-8.61	0.00	--	--	--	--	--	--	--	--	--	--
rs4361660	74041955	0/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs117495229	74044198	G/T	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs188041701	74044269	A/G	0.7	2.4	4.7E-04	0.33	0.17-0.61	0.00	--	--	--	--	--	--	--	--	--	--
rs17145611	74045054	A/G	0.0	0.1	0.58	0.49	0.04-6.36	0.01	--	--	--	--	--	--	--	--	--	--
rs71558537	74050245	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs34762099	74050694	C/T	6.0	6.4	0.36	0.88	0.66-1.16	0.00	--	--	--	--	--	--	--	--	--	--
rs78360862	74056157	C/T	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs35633336	74058292	T/G	5.8	6.5	0.27	0.85	0.64-1.13	0.00	--	--	--	--	--	--	--	--	--	--
rs71539246	74059870	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs13239086	74066052	0/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs35735900	74066227	C/T	5.7	6.2	0.31	0.86	0.64-1.15	0.00	--	--	--	--	--	--	--	--	--	--
rs4717903	74068167	C/T	5.9	6.6	0.25	0.85	0.64-1.13	0.00	--	--	--	--	--	--	--	--	--	--
rs4412258	74068451	G/C	2.4	2.6	0.52	0.87	0.56-1.34	0.01	--	--	--	--	--	--	--	--	--	--
rs13238996	74069645	G/A	5.1	5.5	0.47	0.89	0.66-1.21	0.02	--	--	--	--	--	--	--	--	--	--
rs77800726	74073354	A/G	2.4	2.6	0.45	0.85	0.55-1.31	0.01	--	--	--	--	--	--	--	--	--	--
rs118077281	74076120	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs13238568	74076493	G/A	9.0	10.3	0.12	0.83	0.66-1.05	0.01	--	--	--	--	--	--	--	--	--	--
rs117518423	74077279	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs12670511	74077552	A/G	0.1	0.2	0.21	0.33	0.06-1.88	0.00	--	--	--	--	--	--	--	--	--	--
rs11509861	74082799	0/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs4717905	74083230	A/C	6.0	6.8	0.21	0.83	0.63-1.11	0.01	--	--	--	--	--	--	--	--	--	--
rs117344503	74085363	C/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs6460088	74085759	T/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs200973829	74085812	0/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs4717907	74086493	A/G	5.9	6.8	0.19	0.83	0.62-1.10	0.01	--	--	--	--	--	--	--	--	--	--
rs68008267	74087465	T/C	3.0	1.7	5.1E-03	1.98	1.23-3.19	0.00	--	--	--	--	--	--	--	--	--	--
rs35473599	74088893	A/G	4.4	4.2	0.97	1.01	0.72-1.41	0.02	--	--	--	--	--	--	--	--	--	--
rs36044436	74089819	C/G	5.1	5.5	0.51	0.90	0.66-1.23	0.02	--	--	--	--	--	--	--	--	--	--
rs13227433	74094721	G/T	5.9	6.7	0.28	0.86	0.65-1.14	0.00	--	--	--	--	--	--	--	--	--	--
rs113544043	74095969	A/G	0.0	0.1	0.58	0.49	0.04-6.36	0.01	--	--	--	--	--	--	--	--	--	--
rs3801477	74096481	C/T	1.4	2.7	1.8E-03	0.45	0.28-0.74	0.01	--	--	--	--	--	--	--	--	--	--
rs74531480	74096739	T/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
imm-7-73734951	74097015	0/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs2718271	74097488	T/C	3.1	3.5	0.49	0.88	0.60-1.27	0.01	--	--	--	--	--	--	--	--	--	--
rs56383938	74097622	G/A	0.0	0.1	0.50	0.38	0.02-6.22	0.00	--	--	--	--	--	--	--	--	--	--
rs116861109	74097704	0/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs118009845	74097722	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
imm-7-73736865	74098929	0/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs2527366	74098949	G/A	9.1	10.2	0.19	0.86	0.68-1.08	0.02	--	--	--	--	--	--	--	--	--	--
rs2527367	74099138	C/T	9.1	10.2	0.18	0.86	0.68-1.08	0.02	--	--	--	--	--	--	--	--	--	--
rs6967152	74099317	T/C	5.9	6.5	0.32	0.87	0.65-1.15	0.00	--	--	--	--	--	--	--	--	--	--
rs6959774	74101066	C/T	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs6964833	74101909	C/T	5.9	6.7	0.28	0.86	0.65-1.14	0.00	--	--	--	--	--	--	--	--	--	--
rs186894357	74104658	C/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--	--	--	--	--	--
rs2527380	74104706	T/C	9.0	10.1	0.21	0.86	0.68-1.09	0.02	--	--	--	--	--	--	--	--	--	--
rs35203738	74105196	T/C	5.9	6.4	0.37	0.88	0.66-1.17	0.00	--	--	--	--	--	--	--	--	--	--
rs35275911	74108249	C/G	3.8	2.9	0.09	1.39	0.95-2.05	0.01	--	--	--	--	--	--	--	--	--	--

Immunochip SNP at the *GTF2IRD1-GTF2I* region	Position on Chr7 (hg19)	Allele	Chinese (1010 cases and 848 controls)						European American (930 and 1107)						African American (532 and 367)				
			MAF (%)					r^2 with	MAF (%)						MAF (%)				
			SLE	Control	*P*	OR	95%CI	rs73366469	SLE	Control	*P*	OR	95%CI		SLE	Control	*P*	OR	95%CI
rs79633825	74114342	0/A	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs200579067	74114539	T/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs1064765	74114630	0/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs1057896	74114723	G/T	0.0	0.1	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs2301907	74114847	C/T	5.8	6.7	0.21	0.83	0.63-1.11	0.00	--	--	--	--	--		--	--	--	--	--
rs2259964	74115258	T/C	3.1	3.5	0.45	0.87	0.60-1.26	0.01	--	--	--	--	--		--	--	--	--	--
rs2259965	74115339	T/C	3.1	3.5	0.45	0.87	0.60-1.26	0.01	--	--	--	--	--		--	--	--	--	--
rs185886423	74116329	T/C	0.0	0.1	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs113667755	74118079	C/T	0.1	0.2	0.65	0.67	0.12-3.69	0.00	--	--	--	--	--		--	--	--	--	--
rs78972825	74118351	C/T	0.0	0.1	0.58	0.49	0.04-6.36	0.01	--	--	--	--	--		--	--	--	--	--
rs2301908	74121014	C/T	1.3	2.6	1.6E-03	0.44	0.27-0.73	0.01	--	--	--	--	--		--	--	--	--	--
rs10232099	74121538	A/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs12672029	74122720	A/T	8.9	10.2	0.17	0.85	0.68-1.07	0.02	--	--	--	--	--		--	--	--	--	--
rs7795281	74122854	G/A	5.8	6.7	0.20	0.83	0.63-1.11	0.00	--	--	--	--	--		--	--	--	--	--
rs7795282	74122857	G/A	5.8	6.7	0.20	0.83	0.63-1.11	0.00	--	--	--	--	--		--	--	--	--	--
rs73137172	74126168	T/C	0.1	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs118104623	74127084	G/A	1.3	2.6	1.1E-03	0.43	0.26-0.71	0.01	--	--	--	--	--		--	--	--	--	--
rs116874762	74127959	T/G	1.3	2.7	8.2E-04	0.42	0.25-0.70	0.01	--	--	--	--	--		--	--	--	--	--
rs118014609	74127961	C/A	1.3	2.7	8.3E-04	0.42	0.25-0.70	0.01	--	--	--	--	--		--	--	--	--	--
rs2240367	74128593	A/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs117732260	74129879	A/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
imm-7-73769142	74131206	A/C	0.1	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs17515241	74132014	G/A	5.1	5.5	0.48	0.90	0.66-1.22	0.02	--	--	--	--	--		--	--	--	--	--
rs2718277	74133929	T/C	3.0	3.4	0.26	0.80	0.55-1.18	0.01	--	--	--	--	--		--	--	--	--	--
rs35005436	74134911	C/T	3.6	2.8	0.12	1.36	0.92-2.03	0.01	--	--	--	--	--		--	--	--	--	--
rs6460090	74135844	G/A	0.2	0.6	8.9E-03	0.17	0.05-0.64	0.00	--	--	--	--	--		--	--	--	--	--
rs74818027	74136285	A/G	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs10254080	74137214	0/C	0.0	0.0	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs2267812	74138121	C/A	5.1	5.5	0.44	0.89	0.65-1.21	0.02	--	--	--	--	--		--	--	--	--	--
rs2718278	74138603	T/C	3.1	3.5	0.45	0.87	0.60-1.26	0.01	--	--	--	--	--		--	--	--	--	--
rs2718279	74138940	G/C	3.1	3.5	0.45	0.87	0.60-1.26	0.01	--	--	--	--	--		--	--	--	--	--
rs810364	74140051	C/T	8.2	9.3	0.15	0.83	0.65-1.07	0.03	--	--	--	--	--		--	--	--	--	--
rs76252870	74142220	A/G	0.0	0.1	1.00	--	--	0.00	--	--	--	--	--		--	--	--	--	--
rs707395	74212048	T/C	3.3	3.4	0.66	0.92	0.63-1.34	0.01	--	--	--	--	--		--	--	--	--	--
rs79333834	74256320	A/G	11.7	10.6	0.47	1.08	0.88-1.31	0.01	--	--	--	--	--		--	--	--	--	--
imm-7-73904865	74266929	A/G	3.3	2.5	0.21	1.30	0.86-1.96	0.01	--	--	--	--	--		--	--	--	--	--

Supplementary Table 2. Searching candidate causal SNPs tagged by rs73366469 in the 1000 Genomes Project

1000 Genomes Project SNP	Immunochip SNP	Position on Chr7 (hg38)	Pairwise LD (r^2) with rs73366469 in the 1000 Genomes Project subjects		
			Asian (n=286)	European (n=379)	African (n=246)
rs146236144	No	74580363	0.11	0.00	0.00
rs17145600	No	74580404	0.11	0.00	0.04
rs2267813	No	74582204	0.15	0.05	0.03
rs80346167	No	74586373	0.25	0.27	0.06
rs11981999	No	74587754	0.13	0.18	0.10
rs2005769	No	74590626	0.13	0.05	0.01
rs150108204	No	74594341	0.21	0.13	0.05
rs2019004	No	74595414	0.21	0.21	0.07
rs116657731	No	74597409	0.27	0.29	0.05
rs112806573	No	74598547	0.21	0.34	0.07
rs4717901	No	74602653	0.31	0.06	0.05
rs12667901	No	74607994	**0.73**	**0.40**	**0.31**
rs73366456	No	74608425	**0.74**	**0.40**	**0.39**
rs113066392	No	74611832	0.72	0.63	0.58
rs112502846	No	74611938	0.73	0.71	0.79
rs7800325	No	74618332	0.97	1.00	0.88
rs73366469	Yes	74619286	--	--	--
rs114860935	No	74620209	0.20	0.21	0.14
rs72038845	No	74622249	0.22	0.72	0.42
rs138731063	No	74622806	0.20	0.21	0.14
rs111273641	No	74624371	0.20	0.21	0.11
rs112504285	No	74625217	0.20	0.21	0.12
rs111527316	No	74631507	0.19	0.21	0.10
rs143176121	No	74694107	0.12	0.00	0.00
rs117026326	No	74711703	**0.53**	**0.17**	**0.00**

We hypothesized that Chinese, European Americans and African Americans might share the same causal variant tagged by rs73366469 which should be an untyped functional SNP in strong, modest and weak LD with rs73366469 in Chinese, European Americans and African Americans, respectively. By searching the 1000 Genomes Project phase 1 data set, we found 24 SNPs in LD with rs73366469 in Asians at the level of $r^2 \geq 0.1$ (as shown in the table). None of the 24 SNPs were on the Immunochip. Of these SNPs, rs12667901, rs73366456 and rs117026326 (highlighted in bold and red) were in strong LD with rs73366469 in Asians (defined as $r^2 > 0.5$) but not in Europeans and Africans. We hypothesized these three SNPs as candidate causal variants. To confirm this, rs12667901 and rs117026326 were genotyped using TaqMan assays and assessed for association with SLE. Because rs73366456 was in strong LD with rs12667901 in all three ancestral groups, it was not genotyped.

Supplementary Table 3. Association of p.Arg90His with SLE in Asians

Data set	Chinese-1		All subjects														
			MAF %					*P* after conditioning on								P_{HWE}	
SNP	Position on Chr7 (hg38)	Allele	SLE (n=436)	Control (n=586)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	$\log_{10}$BF	PP	SLE	Control
rs12667901	74607994	T/G	28.2	12.4	1.3E-15	2.90	2.23-3.76	--	2.0E-03	0.50	0.63	1.4E-06	10.9	0.00	0.72	0.01
rs73366469	74619286	C/T	29.7	13.9	1.7E-14	2.66	2.07-3.42	0.86	--	0.54	0.57	1.7E-06	10.0	0.00	0.91	0.02
rs117026326	74711703	T/C	28.1	11.4	1.8E-17	3.18	2.44-4.16	1.9E-03	7.8E-05	--	0.27	1.1E-07	12.6	0.00	0.55	0.01
p.Arg90His	74779296	A/G	38.1	15.6	2.6E-23	3.35	2.64-4.26	1.7E-10	6.0E-12	2.0E-08	--	2.5E-13	18.7	1.00	3.1E-03	0.64
p.Ser99Gly	74779322	G/A	43.2	26.8	1.6E-13	2.22	1.80-2.74	1.1E-03	5.2E-05	0.022	0.70	--	9.6	0.00	0.63	0.83

We measured the ΔGT/GTGT ratio for subjects from different ancestry groups and found the normal ratio of 4:2 in >98% Asian subjects (**Fig. 3c**). Because the impact of mixture of 5:1, 3:3 and 2:4 ratios on association analyses of p.Arg90His in the Asian data sets was negligible (please see the Korean data set as shown below), subjects in the Korean-RA, Chinese-2 and Chinese-SS data sets and some subjects in the Chinese-1 data set were not assessed for the ΔGT/GTGT ratio. In the Chinese-1 data set, 313 cases and 102 controls were measured for the ΔGT/GTGT ratio, of which those with the ratio of 5:1 (2 cases) or 3:3 (3 cases and 3 controls) were excluded for association analyses. Bayes factor (shown as $\log_{10}$BF) was calculated based on the additive model, and posterior probability (shown as PP) was computed for each SNP.

Data set	Chinese-2		All subjects														
			MAF %					*P* after conditioning on								P_{HWE}	
SNP	Position on Chr7 (hg38)	Allele	SLE (n=746)	Control (n=1034)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	$\log_{10}$BF	PP	SLE	Control
rs12667901	74607994	T/G	27.6	13.0	3.9E-21	2.52	2.08-3.05	--	0.11	0.29	0.12	1.1E-08	16.9	0.00	1.00	0.10
rs73366469	74619286	C/T	30.0	15.2	6.5E-22	2.48	2.06-2.99	4.0E-03	--	0.33	0.046	4.3E-11	17.7	0.00	0.49	0.18
rs117026326	74711703	T/C	27.1	11.9	2.3E-24	2.82	2.31-3.44	6.0E-05	2.9E-05	--	0.093	9.0E-11	19.7	0.00	0.17	0.37
p.Arg90His	74779296	A/G	41.6	16.7	1.5E-41	3.27	2.75-3.88	7.7E-27	1.9E-26	2.9E-23	--	1.5E-25	39.2	1.00	3.9E-04	1.00
p.Ser99Gly	74779322	G/A	46.5	28.2	1.6E-22	2.13	1.83-2.48	9.4E-10	2.4E-11	3.7E-08	0.21	--	18.9	0.00	0.42	0.40

Subjects in the Chinese-2 dataset were not measured for the ΔGT/GTGT ratio. Association analyses were carried out in all subjects.

Chinese-1&2 combined	All subjects			
SNP	P_{meta}	OR	$\log_{10}$BF	PP
rs12667901	5.5E-35	2.65	32.9	0.00
rs73366469	9.2E-35	2.55	32.0	0.00
rs117026326	4.4E-40	2.94	37.2	0.00
p.Arg90His	4.2E-63	3.30	63.7	1.00
p.Ser99Gly	2.1E-34	2.16	30.6	0.00

Data set	Korean		Subjects with the ΔGT/GTGT ratio of 4:2														All subjects							
			MAF %					*P* after conditioning on								P_{HWE}	MAF %					*P* after conditioning on		
SNP	Position on Chr7 (hg38)	Allele	SLE (n=611)	Control (n=685)	*P*	OR	95%CI	--	--	rs117026326	p.Arg90His	p.Ser99Gly	$\log_{10}$BF	PP	SLE	Control	SLE (n=614)	Control (n=692)	*P*	OR	95%CI	rs117026326	p.Arg90His	p.Ser99Gly
rs117026326	74711703	T/C	30.0	11.4	3.3E-27	3.25	2.63-4.03	--	--	--	0.18	1.3E-10	23.1	0.0	1.00	0.06	29.9	11.4	3.0E-27	3.24	2.62-4.01	--	0.17	1.2E-10
p.Arg90His	74779296	A/G	46.6	18.1	2.6E-43	3.82	3.16-4.62	--	--	2.2E-22	--	2.3E-25	41.7	1.0	0.52	0.37	46.6	18.1	2.2E-43	3.80	3.14-4.59	1.9E-22	--	2.0E-25
p.Ser99Gly	74779322	G/A	52.4	30.5	3.2E-26	2.47	2.09-2.92	--	--	3.5E-08	0.052	--	22.7	0.0	0.57	0.59	52.4	30.6	2.8E-26	2.46	2.08-2.90	3.0E-08	0.043	--

All Korean subjects were measured for the ΔGT/GTGT ratio.

Association analyses were carried out either in subjects with the normal ΔGT/GTGT ratio of 4:2 (carrying 2 copies each of *NCF1* , *NCF1B* and *NCF1C*) or in all subjects.

Supplementary Table 4. Association of p.Arg90His with SLE in European Americans

Data set EurAm-1

		All subjects										Subjects with the ΔGT/GTGT ratio of 4:2												
		MAF %					*P* after conditioning on					MAF %					*P* after conditioning on							P_{HWE}	
SNP	Allele	SLE (n=716)	Control (n=578)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	SLE (n=612)	Control (n=485)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	$\log_{10}$BF	PP	SLE	Control
rs12667901	T/G	5.4	3.6	0.028	1.56	1.05-2.32	--	0.37	0.57	0.34	0.12	4.9	3.4	0.057	1.55	0.99-2.44	--	0.34	0.78	0.79	0.021	0.2	0.04	0.39	1.00
rs73366469	C/T	11.2	8.3	0.010	1.43	1.09-1.87	0.21	--	0.13	0.22	0.10	10.8	8.5	0.036	1.37	1.02-1.84	0.36	--	0.29	0.64	0.12	0.4	0.05	1.00	0.37
rs117026326	T/C	2.4	0.87	7.2E-03	2.69	1.31-5.55	0.045	0.041	--	0.39	0.036	2.4	0.72	6.4E-03	3.25	1.39-7.56	0.022	0.018	--	0.59	0.019	0.4	0.05	1.00	1.00
p.Arg90His	A/G	8.2	4.1	2.1E-05	2.20	1.53-3.17	2.3E-04	3.6E-04	8.0E-04	--	3.4E-04	5.5	2.1	9.5E-05	2.83	1.68-4.77	8.0E-04	9.5E-04	2.9E-03	--	1.9E-03	1.4	0.61	1.00	1.00
p.Ser99Gly	G/A	17.7	12.5	1.2E-03	1.46	1.16-1.84	6.6E-03	0.014	0.010	0.027	--	17.8	13.0	3.9E-03	1.44	1.12-1.84	1.9E-03	0.016	0.025	0.12	--	1.1	0.25	0.41	0.84

All subjects in the EurAm-1 data set were measured for the ΔGT/GTGT ratio. Association analyses were carried out either in all subjects or in those with the normal ΔGT/GTGT ratio of 4:2.

Data set EurAm-2

		All subjects										Subjects with the ΔGT/GTGT ratio of 4:2												
		MAF %					*P* after conditioning on					MAF %					*P* after conditioning on							P_{HWE}	
SNP	Allele	SLE (n=875)	Control (n=540)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	SLE (n=736)	Control (n=446)	*P*	OR	95%CI	rs12667901	rs73366469	rs117026326	p.Arg90His	p.Ser99Gly	$\log_{10}$BF	PP	SLE	Control
rs12667901	T/G	5.9	4.2	0.040	1.46	1.02-2.11	--	0.46	0.62	0.48	0.55	5.4	3.7	0.035	1.59	1.03-2.45	--	0.50	0.66	0.50	0.52	0.3	0.03	0.26	0.46
rs73366469	C/T	12.0	9.0	0.019	1.37	1.05-1.79	0.25	--	0.20	0.22	0.25	11.9	8.5	0.012	1.46	1.09-1.97	0.15	--	0.16	0.25	0.14	0.7	0.06	1.00	1.00
rs117026326	T/C	2.4	0.74	5.7E-03	3.00	1.38-6.54	0.023	0.020	--	0.16	0.039	2.4	0.67	6.1E-03	3.51	1.43-8.59	0.026	0.023	--	0.29	0.032	0.4	0.03	0.36	1.00
p.Arg90His	A/G	8.3	4.2	3.3E-04	1.92	1.35-2.74	3.7E-03	3.3E-03	0.010	--	0.016	6.0	2.4	5.0E-04	2.42	1.47-3.98	4.2E-03	5.1E-03	0.017	--	0.023	1.3	0.23	0.04	1.00
p.Ser99Gly	G/A	18.2	13.0	3.8E-04	1.50	1.20-1.87	6.8E-03	6.0E-03	4.8E-03	0.026	--	18.6	13.1	5.5E-04	1.53	1.20-1.95	0.011	6.9E-03	6.6E-03	0.057	--	1.7	0.65	0.18	0.21

All subjects in the EurAm-2 data set were measured for the ΔGT/GTGT ratio. Association analyses were carried out either in all subjects or in those with the normal ΔGT/GTGT ratio of 4:2.

EurAm-1&2 combined

	All subjects		Subjects with the ΔGT/GTGT ratio of 4:2			
SNP	P_{meta}	OR	P_{meta}	OR	$\log_{10}$BF	PP
rs12667901	2.7E-03	1.51	4.5E-03	1.57	0.9	0.00
rs73366469	5.0E-04	1.40	1.1E-03	1.42	1.6	0.00
rs117026326	1.2E-04	2.83	1.1E-04	3.37	1.4	0.00
p.Arg90His	3.0E-08	2.05	1.9E-07	2.61	4.1	0.83
p.Ser99Gly	1.5E-06	1.48	7.3E-06	1.49	3.4	0.17

Supplementary Table 5. Association of p.Arg90His with SLE in African Americans

Data set AfrAm			**All subjects**					**Subjects with the ΔGT/GTGT ratio of 4:2**								
			MAF %					MAF %							P_{HWE}	
SNP	Position on Chr7 (hg38)	Allele	SLE (n=532)	Control (n=367)	*P*	OR	95%CI	SLE (n=474)	Control (n=315)	*P*	OR	95%CI	log_{10}BF	PP	SLE	Control
rs73366469	74619286	C/T	17.7	19.6	0.32	0.88	0.69-1.13	18.5	19.5	0.66	0.94	0.73-1.23	-0.2	0.00	0.44	0.86
rs117026326[a]	74711703	T/C	0.39	0.0	1.00	--	--	0.43	0.0	1.00	--	--	--	--	1.00	1.00
p.Arg90His	74779296	A/G	16.8	10.1	7.0E-05	1.80	1.35-2.40	15.7	8.3	2.9E-05	2.02	1.45-2.81	2.4	1.00	0.30	0.15
p.Ser99Gly	74779322	G/A	40.0	39.7	0.88	1.02	0.83-1.24	40.1	39.4	0.78	1.03	0.83-1.27	-0.3	0.00	0.45	0.41
Intronic-1[a]	74780073	C/G	0.39	0.0	1.00	--	--	0.43	0.0	1.00	--	--	--	--	1.00	1.00
Intronic-2[a]	74781840	A/C	0.39	0.0	1.00	--	--	0.43	0.0	1.00	--	--	--	--	1.00	1.00

All subjects in the AfrAm data set were measured for the ΔGT/GTGT ratio.

Association analyses were carried out either in all subjects or in those with the normal ΔGT/GTGT ratio of 4:2.

[a]rs117026326, intronic-1 and intronic-2 were genotyped in a subset of African-American subjects (382 cases and 227 controls).

Supplementary Table 6. Genotype of *NCF1* variants identified in Chinese subjects carrying risk and non-risk genotypes of rs117026326 by Sanger sequencing

dbSNP ID	Position on Chr7 (hg38)	Location on *NCF1*	Variant on *NCF1*	r^2 with rs117026326	Subjects (n=23) carrying the SLE-risk TT genotype of rs117026326																							Subjects (n=22) carrying the non-risk CC genotype of rs117026326																						Reference allele at *NCF1/1B/1C*
					1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	23	1	2	3	4	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21	22	
rs117026326					TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	
	74774752	Intron1	G/T	0.07	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GT	GG	GT	GG	GT	GT	GT	GT	GG	GG	GG	GG	GG	GG	GG	GG	GT	GG	GG	T/T/T
	74774887	Intron1	C/T	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CT	C/C/C
	74775104	Intron1	C/A	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	C/C/C
	74775655	Intron1	C/G	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CG	CC	CC	CC	C/C/C
rs140034807	74775881	Intron1	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
rs138054188	74775882	Intron1	C/T	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/C
rs2906281	74776039	Intron1	C/T	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	C/C/C
rs781939751	74776509	Intron1	C/A	0.05	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	CC	CC	AC	AC	AC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	A/C/C
	74776716	Intron1	G/T	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GT	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
	74776717	Intron1	T/C	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	T/T/T
	74777279	Exon2	C/T (p.Leu29Leu)	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/C
rs145170670	74777361	Intron2	C/T	0.05	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CT	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	T/C/C
	74778377	Intron2	C/T	0.03	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CT	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/T/T
	74778595	Intron2	G/A	0.03	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	AG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/A
rs201625983	74778644	Intron2	C/T	0.03	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CT	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/T/T
rs200623471	74778736	Intron2	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	A/G/G
	74778776	Intron2	G/A	0.03	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	AG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/A/A
rs368231459	74779020	Intron2	C/A	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	C/C/C
rs782395518	74779138	Exon3	G/C (p.Arg70Ser)	0.02	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	CG	G/G/G
	74779221	Intron3	C/A	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/A/C
rs201802880	74779296	Exon4	G/A (**p.Arg90His**)	**0.67**	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AG	AG	AG	AG	AG	AG	AG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/A/A
rs17295741	74779322	Exon4	A/G (**p.Ser99Gly**)	**0.73**	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	AG	AG	AG	AG	AG	AA	AA	AA	AG	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	A/G/A
rs17356100	74779372	Exon4	C/T (p.Leu115Leu)	0.03	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CT	C/T/C
rs587670410	74779456	Intron4	G/- deletion	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G-	GG	GG	GG	GG	GG	GG	GG	G/G/G
	74780073	Intron4	G/C (**intronic-1**)	**0.87**	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CG	CC	CC	CC	CC	CC	CC	CC	CC	CC	CG	CC	CC	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
rs782512803	74780860	Intron5	-/G insertion	0.01	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	G-	--	--	-/-/-
rs782366672	74781407	Intron5	T/C	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	T/C/C
rs800977	74781492	Intron5	C/T	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	C/C/C
rs782657204	74781580	Intron5	T/C	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	T/C/C
	74781655	Intron5	C/T	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/T
rs140969778	74781714	Intron5	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/A/A
rs800978	74781743	Intron5	G/A	0.02	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	A/G/G
rs376392702	74781748	Intron5	-/T insertion	0.01	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	--	T-	--	--	-/-/-
rs800979	74781755	Intron5	A/G	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	AA	AA	AA	AG	AA	AA	AA	AA	AA	AG	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	AA	G/G/G
rs371061025	74781786	Intron5	G/C	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
rs199789198	74781840	Intron5	C/A (**intronic-2**)	**0.91**	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AC	AA	AA	AA	CC	CC	CC	CC	CC	AC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/C
rs800980	74781899	Intron5	C/T	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	T/C/C
rs2523327	74781945	Intron5	C/T	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/T/T
	74781952	Intron5	A/G	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	A/G/G
rs190079672	74782003	Intron5	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/A/A
rs800981	74782019	Intron5	C/T	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	T/C/C
	74782482	Intron5	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	A/G/A
rs782555266	74782983	Exon6	A/G (p.Asn166Asp)	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	AA	A/A/G
rs62475426	74783255	Intron6	A/T	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AT	AA	A/A/A
rs2528941	74783387	Intron6	A/G	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	A/A/A
	74784077	Intron7	T/C	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	T/T/T
rs147472798	74784109	Intron7	T/C	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	CC	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	C/C/C
	74784815	Intron7	G/C	0.02	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	C/C/C
rs369485834	74784878	Intron7	G/A	0.02	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
	74784905	Intron7	A/G	0.01	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AG	AA	AA	AA	A/A/A
	74785049	Intron7	G/C	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	G/G/G
rs191081238	74785092	Intron7	C/G	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CG	CC	CC	C/C/C
rs373252492	74786173	Intron8	A/T	0.03	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AT	AA	AA	AA	AA	AA	AA	TT	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	T/T/T
	74786613	Intron8	C/T	0.01	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/C
rs200877252	74786994	Intron8	T/G	0.01	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	GT	TT	TT	T/T/T
rs149863211	74787254	Intron8	T/C	0.02	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	C/C/C
	74787843	Intron8	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	G/G/G
rs2539044	74788173	Intron9	G/A	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	AG	GG	GG	G/G/G
	74788320	Intron9	C/G	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CG	CC	CC	CC	CC	CC	CC	CG	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	G/C/C
rs587631811	74788699	Exon10	C/A (p.Pro349Gln)	0.01	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	AC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	C/C/C
rs587619282	74788740	Intron10	G/C	0.03	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	GG	CG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	C/C/C
rs4717143	74788777	Intron10	T/C	0.02	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	CT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	T/T/C
	74788845	Intron10	T/G	0.00	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	GT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	TT	T/G/G
	74788952	Intron10	C/A	0.02	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AC	AA	AA	AA	AA	AA	AA	AC	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	AA	C/C/A
	74788996	Intron10	T/G	0.00	GG	GG	TT	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GT	GG	GG	GG	GG	GG	GG	GT	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	T/T/T
	74789048	Exon11	G/T (p.Arg354Leu)	0.01	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GG	GT	GG	GG	G/G/G
	74789215	3'UTR	T/C	0.02	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CT	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	CC	T/T/C

Reference alleles at *NCF1* , *NCF1B* and *NCF1C* are based on the human genome build GRCh38/hg38. The dbSNP ID is shown if available. SNPs in LD with rs117026326 are highlighted by a blue background. The minor allele is highlighted in red.
All 45 subjects showed the normal ΔGT/GTGT ratio of 4:2.

Supplementary Table 7. Association of p.Arg90His with primary Sjögren's syndrome in European Americans and Chinese

Data set	EurAm-SS		Subjects with the ΔGT/GTGT ratio of 4:2												All subjects							
			MAF %					*P* after conditioning on					P_{HWE}		MAF %					*P* after conditioning on		
SNP	Position on Chr7 (hg38)	Allele	SS (n=302)	Control (n=931)	*P*	OR	95%CI	rs117026326	p.Arg90His	p.Ser99Gly	log_{10}BF	PP	SS	Control	SS (n=373)	Control (n=1118)	*P*	OR	95%CI	rs117026326	p.Arg90His	p.Ser99Gly
rs117026326	74711703	T/C	2.0	0.70	7.3E-03	3.12	1.36-7.17	--	0.36	0.017	0.2	0.21	1.00	1.00	1.9	0.81	0.018	2.43	1.16-5.09	--	0.28	0.042
p.Arg90His	74779296	A/G	4.8	2.2	9.7E-04	2.35	1.42-3.91	0.036	--	4.0E-03	0.7	0.66	0.51	1.00	7.0	4.1	2.8E-03	1.75	1.21-2.53	0.038	--	0.016
p.Ser99Gly	74779322	G/A	15.4	13.1	0.20	1.19	0.91-1.55	0.54	0.927	--	0.0	0.13	0.51	0.56	15.0	12.8	0.19	1.18	0.92-1.50	0.50	0.50	--

All subjects in the EurAm-SS data set were measured for the ΔGT/GTGT ratio. Association analyses were carried out either in all subjects or in those with the normal ΔGT/GTGT ratio of 4:2.

Data set	Chinese-SS		All subjects											
			MAF %					*P* after conditioning on					P_{HWE}	
SNP	Position on Chr7 (hg38)	Allele	SS (n=449)	Control (n=469)	*P*	OR	95%CI	rs117026326	p.Arg90His	p.Ser99Gly	log_{10}BF	PP	SS	Control
rs117026326	74711703	T/C	26.2	14.7	4.6E-09	2.03	1.60-2.57	--	0.29	1.9E-03	5.6	0.00	0.71	0.71
p.Arg90His	74779296	A/G	37.8	18.3	7.2E-17	2.45	1.98-3.02	4.3E-10	--	7.3E-13	13.0	1.00	1.2E-03	0.35
p.Ser99Gly	74779322	G/A	41.5	29.1	9.7E-08	1.68	1.39-2.04	0.064	0.017	--	4.9	0.00	0.02	0.74

Subjects in the Chinese-SS data set were not measured for the ΔGT/GTGT ratio. Association analyses were carried out in all subjects.

Supplementary Table 8. Independent association of p.Arg90His with seropositive rheumatoid arthritis in Koreans

Data set	**Korean-RA**		MAF %					*P* afte conditioning on				P_{HWE}	
SNP	Position on Chr7 (hg38)	Allele	RA (n=863)	Control (n=685)	*P*	OR	95%CI	p.Arg90His	p.Ser99Gly	log_{10}BF	PP	RA	Control
p.Arg90His	74779296	A/G	26.6	18.1	2.5E-08	1.65	1.38-1.97	--	1.1E-05	5.5	1.00	0.30	0.45
p.Ser99Gly	74779322	G/A	35.2	30.5	2.0E-03	1.27	1.09-1.49	0.94	--	1.4	0.00	0.55	0.93

Korean patients with rheumatoid arthritis were not measured for the ΔGT/GTGT ratio.

Korean controls were measured for the ΔGT/GTGT ratio, and only those with the 4:2 ratio were included in associaton analyses.

Supplementary Table 9. Association of copy number variation of *NCF1* with SLE and SS

				5:1 ratio					3:3 and 2:4 ratio					4:2 ratio (reference)	
		Subject		N[a] (frequency%)					N[a] (frequency%)					N[a] (frequency%)	
Disease	Ancestry	Case	Control	Case	Control	*P*	OR	95% CI	Case	Control	*P*	OR	95% CI	Case	Control
SLE	Korean	829	782	3 (0.4)	0 (0.0)	0.032	-	-	0 (0.0)	7 (0.9)	3.7E-05	--	--	826 (99.6)	775 (99.1)
SLE	Chinese	511	573	3 (0.6)	0 (0.0)	0.011	-	-	3 (0.6)	12 (2.1)	2.8E-03	0.28	0.11-0.68	505 (98.8)	561 (97.9)
SLE	EurAm	1593	1118	17 (1.1)	3 (0.3)	5.9E-04	3.91	1.64-9.33	227 (14.2)	184 (16.5)	0.038	0.85	0.73-0.99	1349 (84.7)	931 (83.3)
SLE	AfrAm	716	406	4 (0.6)	1 (0.2)	0.509	2.19	0.46-10.4	73 (10.2)	55 (13.5)	0.018	0.73	0.56-0.95	639 (89.2)	350 (86.2)
SS	EurAm	382	1118	3 (0.8)	3 (0.3)	0.085	3.02	0.97-9.41	71 (18.6)	184 (16.5)	0.163	1.17	0.94-1.45	308 (80.6)	931 (83.3)

[a]number

Supplementary Table 10. *NCF1* -specific PCR and sequencing primers

PCR product	PCR primer	Sequencing primer
P1 (3375bp)	P1-L (forward): 5'-GCAAAGCCCTTTCTGTACCC P1-R (reverse): 5'-TCACCAGGAACATGTACACCTG	P1-1: 5'-GCAAAGCCCTTTCTGTACCC P1-2: 5'-AGGCCAGGGAGGAACTACAT P1-3: 5'-CGTGAGCTACCGTGCCTAGT P1-4: 5'-ATCCACTGCCCTGGATACAC P1-5: 5'-CGGATTCTGGATCGGTCTTA P1-6: 5'-TCACCAGGAACATGTACACCTG P1-7: 5'-GTTTGTGCCCTTTCTGCAAT
P2 (6466bp)	P2-L (forward): 5'-GCTTTCCCCCAGGTGTACA P2-R (reverse): 5'-CCCAGACTCCAAAGCAGTCA	P2-1: 5'-AAAGCGACAAGAATGGTTCC P2-2: 5'-TAACGAAAATGGCCTCCTTG P2-3: 5'-CCTTGTGGTTCTCGTCCCTA P2-4: 5'-AGCAAAAGGATTTGCTGGTG P2-5: 5'-CTGTCAGGGGGTCATTGG P2-6: 5'-TGTCTGGGGTCTCCTGTACC P2-7: 5'-CCTGAACACTCTGGGTCTCC P2-8: 5'-TGGGAAGTTCTTCTGCAGGT P2-9: 5'-TTTCTGACCAGCCCTTCATC P2-10: 5'-AGGGCCCACTCTACTGACCT P2-11: 5'-TTGTGGCTGTGGGTTCCT P2-12: 5'-CCCAGACTCCAAAGCAGTCA
P3 (6298bp)	P3-L (forward): 5'-AAGGCTCAGGCAGCCTTG P3-R (reverse): 5'-CAGACCTCGGCTCTTGCAACTT	P3-1: 5'-GTGTCCTCACCATCCCTGAT P3-2: 5'-TTACAAGCATGCACCACCAT P3-3: 5'-TCTACCTGGCACTGGGAACT P3-4: 5'-ACCCCAGATCCCTAAACACC P3-5: 5'-AGGCGCAGAGAGGAGAAGAT P3-6: 5'-TGGGGTCAAGTGATCCTTTC P3-7: 5'-TCATGCCTGTAATCCCAACA P3-8: 5'-CTCCAGCTTGGGAGTCAGAG P3-9: 5'-TTTCTGGTTCTGGGGAACAC P3-10: 5'-TCCTGTCTCAGCTTCCCAAG P3-11: 5'-ATGGCCTAGACAGAGCGAAC P3-12: 5'-AGAGAGCCCTGAAACCCTCT P3-13: 5'-CCCAGCAGAACTCACAACCT
P2* (2208bp)	P2-L* (forward): 5'-GCTTTCCCCCAGGTGTAC P2-R* (reverse): 5'-CCTTCCCTCTCCCACCT	

Supplementary Table 11. TaqMan assays for SNP genotyping and the ΔGT/GTGT ratio

Target	Forward Primer Sequence	Reverse Primer Sequence	Reporter 1 Sequence	Reporter 2 Sequence
rs12667901	TGGTGACGGATGCCTGTAATC	ACCTTCTGGTTTCAAGCGATTCTC	CTGCCTCAGCCTCCCGA	CTGCCTCAGACTCCCGA
rs73366469	CGTGTGCCTGAGCAATTGAG	ACAGCCTGGCCCTCTCT	CTTCCTCCTGCAGGCC	TCCTCCCGCAGGCC
rs117026326	GGTTAGTTTGCATTTTCTATAAAGTCTTATGAATGAAATA	GCTGTGGATGAATTTCAAAACAATCATTT	CTCCCCGGCCCATG	CTCCCCAGCCCATG
p.Arg90His	CAGCTCCCAAGTGGTTTGAC	GGTGGGCAGGCTCATGA	CCTGGCGGTTCTC	CCTGGTGGTTCTC
p.Ser99Gly	CGGGCCGCCGAGAA	TGGTGGGCAGGCTCATG	AGTACTGCGGCACGCT	AGTACTGCAGCACGCT
Intronic-1	AGCACTTATTGACGCTTGAAGGT	GCCTCCCGGGCTGATG	CTGCCCTTCCTTGGGC	TGCCCTTCGTTGGGC
Intronic-2	CCTCCCAAAGTGCTGAGATTATAGA	CAACAGGCCTCGTGTCCTTA	CCGGCTTTTTCTTCTTCTTA	CGGCTTTTTCTTATTCTTA
GTGT	CCCCGACTCTGGCTTTCC	CCGACAGGTCCTGCCATTT	CCCAGGTGTACATGTT	
ΔGT	CCCCGACTCTGGCTTTCC	CCGACAGGTCCTGCCATTT	CCCAGGTACATGTTCCT	