

Supplementary data table S-1: Genes Implicated in Systemic Lupus Erythematosus.

Symbol	Chromosome	SLE	LN	Caucasians	African American	Asian	Hispanic	CNV	epigenetics	miRNA
ATG5	6	YES [1][2]		YES [1][3]		YES [4]				
ACTN4	19	YES [5]		YES [5]						
BANK1	4	YES [6]		YES [6]						
BLK	8	YES [1][7][8]		YES [7] [1][8][3]		YES [4][9]				
C1QA	1	YES [10][11][12]	YES [13]							
C1QB	1	YES [10][11][12]	YES [13]							
C2	6	YES [10][11][12]								
C4A	6	YES [10][11][12] [14]		YES [14]				YES [14] (caucasians)		
C4B	6	YES [10][11][12] [14]		YES [14]				YES [14] (caucasians)		
ATF6B (CREBL1)	6	YES [15]								
CRP	1	YES [16]		YES [17][18][19]	YES [17][18][19]					
CD44	11	YES [5]		YES [5]						
DNAJA1	9	YES [5]		YES [5]						

ETS1	11					YES [4]'[9]				
FCGR1A	1	YES [16]								
FCGR1B	1	YES [16]								
FCGR2A	1	YES [16]	YES [20]	YES [1]						
FCGR2B	1	YES [16]								
FCGR3A	1	YES [21]	YES [22]'[20]		YES [23]		YES LN [22]			
FCGR3B	1	YES [21]'[24]	YES glomerulo- nephritis [25]			YES [22]'[26]		YES [25]' [27]'[28]		
FRK-PTPRD pair	6; 9	YES [5]		YES [5]						
HLA-DRB1		YES [15]		YES [3]'[15]		YES [4]'[9]				
HLA-DRB2	6	YES [29]	NO [30]'[31]	YES [29]	NO [29]	YES [4]'[9]				
HLA-DRB3	6	YES [29]	NO [30]'[31]	YES [29]'[7]	NO [29]	YES [4]'[9]				
ICA1	7	[1]								
IKZF1	7			YES [3]		YES [4]'				
IL10	1			YES [1]'[3]						
IL21	4	YES [32]		YES [32]	YES [32]					
IRAK1 (MECP2)	X	YES [33]		YES [3]						YES mir146 [34]
IRF5	7	YES [35, 36]		YES [1]'[3]		YES [4]'[9]				YES mir146 [34]

IRF53 (not determined)				YES [7]' [8]						
IRF8	16	YES [5]		YES [5]						
ITGAM	16	YES [1]'[7]'[37]		YES [38]' [7]' [1]' [8]'[3]	YES [38]	NO [38]				
JAZF1	7			YES [1]'[3]						
KPNA1	3	YES [5]		YES [5]						
LRRC18 (WDFY4)						YES [4]'[9]				
Lyn				YES [1]						
MECP2	X	YES [39]'[40]'[41]								
MICB	6	YES [15]		YES [15]						
NMNAT2	1	YES [1]								
NOTCH4	6	YES [42]								
OR2H2	6	YES [15]		YES [15]						
PDCD1	2	YES [43]	YES [44]'[45]'[46]' [47]							

STAT4	2	YES [53]	YES [54]	YES [7]' YES [1]' [8]' [3]		YES [4]' [9]				
SYK	9	YES [5]		YES [5]						
TAP2	6	YES [55]								
TNFAIP3	6	YES [8]		YES [8]'[3]		YES [4]' [9]				
TNFSF4	1	YES [56][57]		YES [3]		YES [4]'				
TNIP1	5			YES [1]'[3]		YES [4]'				
TRAF6	11									YES mir146 [34]
TREX1	3	YES [58]								
TRIM27	6	YES [15]		YES [15]						
TRIM31	6	YES [42]								
TYK2	19	YES [35]'[39]'[3]								
UBE2L3 (HIC)	22	YES [1]'[2]'[5]		YES [1]' [3]		YES [4]				
UHRF1BP1	6			YES [1]' [3]						
XKR6				YES [1]						

References

1. Harley JB, Alarcon-Riquelme ME, Criswell LA, et al. Genome-wide association scan in women with systemic lupus erythematosus identifies susceptibility variants in ITGAM, PXK, KIAA1542 and other loci. *Nat Genet* 2008;40(2):204-10.
2. Graham RR, Hom G, Ortmann W, Behrens TW. Review of recent genome-wide association scans in lupus. *J Intern Med* 2009;265(6):680-8.
3. Gateva V, Sandling JK, Hom G, et al. A large-scale replication study identifies TNIP1, PRDM1, JAZF1, UHRF1BP1 and IL10 as risk loci for systemic lupus erythematosus. *Nat Genet* 2009;41(11):1228-33.
4. Han JW, Zheng HF, Cui Y, et al. Genome-wide association study in a Chinese Han population identifies nine new susceptibility loci for systemic lupus erythematosus. *Nat Genet* 2009;41(11):1234-7.
5. Ramos PS, Williams AH, Ziegler JT, et al. Genetic analyses of interferon pathway-related genes reveals multiple new loci associated with systemic lupus erythematosus (SLE). *Arthritis Rheum* 2011.
6. Kozyrev SV, Abelson AK, Wojcik J, et al. Functional variants in the B-cell gene BANK1 are associated with systemic lupus erythematosus. *Nat Genet* 2008;40(2):211-6.
7. Hom G, Graham RR, Modrek B, et al. Association of systemic lupus erythematosus with C8orf13-BLK and ITGAM-ITGAX. *N Engl J Med* 2008;358(9):900-9.
8. Graham RR, Cotsapas C, Davies L, et al. Genetic variants near TNFAIP3 on 6q23 are associated with systemic lupus erythematosus. *Nat Genet* 2008;40(9):1059-61.
9. Yang W, Shen N, Ye DQ, et al. Genome-wide association study in Asian populations identifies variants in ETS1 and WDFY4 associated with systemic lupus erythematosus. *PLoS Genet* 2010;6(2):e1000841.
10. Agnello V, De Bracco MM, Kunkel HG. Hereditary C2 deficiency with some manifestations of systemic lupus erythematosus. *J Immunol* 1972;108(3):837-40.
11. Hauptmann G, Grosshans E, Heid E. Lupus erythematosus syndrome and complete deficiency of the fourth component of complement. *Boll Ist Sieroter Milan* 1974;53(1):suppl:228.
12. Nishino H, Shibuya K, Nishida Y, Mushimoto M. Lupus erythematosus-like syndrome with selective complete deficiency of C1q. *Ann Intern Med* 1981;95(3):322-4.
13. Pickering MC, Botto M, Taylor PR, Lachmann PJ, Walport MJ. Systemic lupus erythematosus, complement deficiency, and apoptosis. *Adv Immunol* 2000;76:227-324.

14. Yang Y, Chung EK, Wu YL, et al. Gene copy-number variation and associated polymorphisms of complement component C4 in human systemic lupus erythematosus (SLE): low copy number is a risk factor for and high copy number is a protective factor against SLE susceptibility in European Americans. *Am J Hum Genet* 2007;80(6):1037-54.
15. Barcellos LF, May SL, Ramsay PP, et al. High-density SNP screening of the major histocompatibility complex in systemic lupus erythematosus demonstrates strong evidence for independent susceptibility regions. *PLoS Genet* 2009;5(10):e1000696.
16. Brown EE, Edberg JC, Kimberly RP. Fc receptor genes and the systemic lupus erythematosus diathesis. *Autoimmunity* 2007;40(8):567-81.
17. Edberg JC, Wu J, Langefeld CD, et al. Genetic variation in the CRP promoter: association with systemic lupus erythematosus. *Hum Mol Genet* 2008;17(8):1147-55.
18. Jonsen A, Gunnarsson I, Gullstrand B, et al. Association between SLE nephritis and polymorphic variants of the CRP and FcgammaRIIIa genes. *Rheumatology (Oxford)* 2007;46(9):1417-21.
19. Russell AI, Cunningham Graham DS, Shepherd C, et al. Polymorphism at the C-reactive protein locus influences gene expression and predisposes to systemic lupus erythematosus. *Hum Mol Genet* 2004;13(1):137-47.
20. Karassa FB, Trikalinos TA, Ioannidis JP. The Fc gamma RIIIA-F158 allele is a risk factor for the development of lupus nephritis: a meta-analysis. *Kidney Int* 2003;63(4):1475-82.
21. Wu J, Edberg JC, Redecha PB, et al. A novel polymorphism of FcgammaRIIIa (CD16) alters receptor function and predisposes to autoimmune disease. *J Clin Invest* 1997;100(5):1059-70.
22. Kyogoku C, Dijstelbloem HM, Tsuchiya N, et al. Fcgamma receptor gene polymorphisms in Japanese patients with systemic lupus erythematosus: contribution of FCGR2B to genetic susceptibility. *Arthritis Rheum* 2002;46(5):1242-54.
23. Salmon JE, Millard S, Schachter LA, et al. Fc gamma RIIA alleles are heritable risk factors for lupus nephritis in African Americans. *J Clin Invest* 1996;97(5):1348-54.
24. Ptacek T, Li X, Kelley JM, Edberg JC. Copy number variants in genetic susceptibility and severity of systemic lupus erythematosus. *Cytogenet Genome Res* 2008;123(1-4):142-7.
25. Aitman TJ, Dong R, Vyse TJ, et al. Copy number polymorphism in Fcgr3 predisposes to glomerulonephritis in rats and humans. *Nature* 2006;439(7078):851-5.
26. Lee YH, Ji JD, Song GG. Fcgamma receptor IIB and IIIB polymorphisms and susceptibility to systemic lupus erythematosus and lupus nephritis: a meta-analysis. *Lupus* 2009;18(8):727-34.
27. Fanciulli M, Norsworthy PJ, Petretto E, et al. FCGR3B copy number variation is associated with susceptibility to systemic, but not organ-specific, autoimmunity. *Nat Genet* 2007;39(6):721-3.
28. Willcocks LC, Lyons PA, Clatworthy MR, et al. Copy number of FCGR3B, which is associated with systemic lupus erythematosus, correlates with protein expression and immune complex uptake. *J Exp Med* 2008;205(7):1573-82.

29. Tsao BP, Wu H. The genetics of human lupus. In: Wallace DJ, Hahn BH, editors. *Dubois' lupus erythematosus*. Philadelphia: Lippincott Williams & Wilkins; 2007. p. 54-81.
30. Freedman BI, Spray BJ, Heise ER, Espeland MA, Canzanello VJ. A race-controlled human leukocyte antigen frequency analysis in lupus nephritis. The South-Eastern Organ Procurement Foundation. *Am J Kidney Dis* 1993;21(4):378-82.
31. Harley JB, Sestak AL, Willis LG, Fu SM, Hansen JA, Reichlin M. A model for disease heterogeneity in systemic lupus erythematosus. Relationships between histocompatibility antigens, autoantibodies, and lymphopenia or renal disease. *Arthritis Rheum* 1989;32(7):826-36.
32. Hughes T, Kim-Howard X, Kelly JA, et al. Fine-mapping and transethnic genotyping establish IL2/IL21 genetic association with lupus and localize this genetic effect to IL21. *Arthritis Rheum* 2011;63(6):1689-97.
33. Jacob CO, Reiff A, Armstrong DL, et al. Identification of novel susceptibility genes in childhood-onset systemic lupus erythematosus using a uniquely designed candidate gene pathway platform. *Arthritis Rheum* 2007;56(12):4164-73.
34. Tang Y, Luo X, Cui H, et al. MicroRNA-146A contributes to abnormal activation of the type I interferon pathway in human lupus by targeting the key signaling proteins. *Arthritis Rheum* 2009;60(4):1065-75.
35. Sigurdsson S, Nordmark G, Goring HH, et al. Polymorphisms in the tyrosine kinase 2 and interferon regulatory factor 5 genes are associated with systemic lupus erythematosus. *Am J Hum Genet* 2005;76(3):528-37.
36. Kozyrev SV, Alarcon-Riquelme ME. The genetics and biology of Irf5-mediated signaling in lupus. *Autoimmunity* 2007;40(8):591-601.
37. Nath SK, Han S, Kim-Howard X, et al. A nonsynonymous functional variant in integrin-alpha(M) (encoded by ITGAM) is associated with systemic lupus erythematosus. *Nat Genet* 2008;40(2):152-4.
38. Han S, Kim-Howard X, Deshmukh H, et al. Evaluation of imputation-based association in and around the integrin-alpha-M (ITGAM) gene and replication of robust association between a non-synonymous functional variant within ITGAM and systemic lupus erythematosus (SLE). *Hum Mol Genet* 2009;18(6):1171-80.
39. Suarez-Gestal M, Calaza M, Endreffy E, et al. Replication of recently identified systemic lupus erythematosus genetic associations: a case-control study. *Arthritis Res Ther* 2009;11(3):R69.
40. Sawalha AH, Webb R, Han S, et al. Common variants within MECP2 confer risk of systemic lupus erythematosus. *PLoS One* 2008;3(3):e1727.
41. Webb R, Wren JD, Jeffries M, et al. Variants within MECP2, a key transcription regulator, are associated with increased susceptibility to lupus and differential gene expression in patients with systemic lupus erythematosus. *Arthritis Rheum* 2009;60(4):1076-84.
42. Rioux JD, Goyette P, Vyse TJ, et al. Mapping of multiple susceptibility variants within the MHC region for 7 immune-mediated diseases. *Proc Natl Acad Sci U S A* 2009;106(44):18680-5.
43. Prokunina L, Castillejo-Lopez C, Oberg F, et al. A regulatory polymorphism in PDCD1 is associated with susceptibility to systemic lupus erythematosus in humans. *Nat Genet* 2002;32(4):666-9.
44. Johansson M, Arlestig L, Moller B, Rantapaa-Dahlqvist S. Association of a PDCD1 polymorphism with renal manifestations in systemic lupus erythematosus. *Arthritis Rheum* 2005;52(6):1665-9.

45. Nielsen C, Lastrup H, Voss A, Junker P, Husby S, Lillevang ST. A putative regulatory polymorphism in PD-1 is associated with nephropathy in a population-based cohort of systemic lupus erythematosus patients. *Lupus* 2004;13(7):510-6.
46. Prokunina L, Gunnarsson I, Sturfelt G, et al. The systemic lupus erythematosus-associated PDCD1 polymorphism PD1.3A in lupus nephritis. *Arthritis Rheum* 2004;50(1):327-8.
47. Lee YH, Woo JH, Choi SJ, Ji JD, Song GG. Association of programmed cell death 1 polymorphisms and systemic lupus erythematosus: a meta-analysis. *Lupus* 2009;18(1):9-15.
48. Tan W, Sunahori K, Zhao J, et al. Association of PPP2CA polymorphisms with SLE susceptibility in multiple ethnic groups. *Arthritis Rheum* 2011.
49. Kyogoku C, Langefeld CD, Ortmann WA, et al. Genetic association of the R620W polymorphism of protein tyrosine phosphatase PTPN22 with human SLE. *Am J Hum Genet* 2004;75(3):504-7.
50. Wu H, Cantor RM, Graham DS, et al. Association analysis of the R620W polymorphism of protein tyrosine phosphatase PTPN22 in systemic lupus erythematosus families: increased T allele frequency in systemic lupus erythematosus patients with autoimmune thyroid disease. *Arthritis Rheum* 2005;52(8):2396-402.
51. Fernando MM, Stevens CR, Sabeti PC, et al. Identification of two independent risk factors for lupus within the MHC in United Kingdom families. *PLoS Genet* 2007;3(11):e192.
52. Moser KL, Kelly JA, Lessard CJ, Harley JB. Recent insights into the genetic basis of systemic lupus erythematosus. *Genes Immun* 2009;10(5):373-9.
53. Remmers EF, Plenge RM, Lee AT, et al. STAT4 and the risk of rheumatoid arthritis and systemic lupus erythematosus. *N Engl J Med* 2007;357(10):977-86.
54. Taylor KE, Remmers EF, Lee AT, et al. Specificity of the STAT4 genetic association for severe disease manifestations of systemic lupus erythematosus. *PLoS Genet* 2008;4(5):e1000084.
55. Ramos PS, Langefeld CD, Bera LA, Gaffney PM, Noble JA, Moser KL. Variation in the ATP-binding cassette transporter 2 gene is a separate risk factor for systemic lupus erythematosus within the MHC. *Genes Immun* 2009;10(4):350-5.
56. Cunningham Graham DS, Graham RR, Manku H, et al. Polymorphism at the TNF superfamily gene TNFSF4 confers susceptibility to systemic lupus erythematosus. *Nat Genet* 2008;40(1):83-9.
57. Delgado-Vega AM, Abelson AK, Sanchez E, et al. Replication of the TNFSF4 (OX40L) promoter region association with systemic lupus erythematosus. *Genes Immun* 2009;10(3):248-53.
58. Lee-Kirsch MA, Gong M, Chowdhury D, et al. Mutations in the gene encoding the 3'-5' DNA exonuclease TREX1 are associated with systemic lupus erythematosus. *Nat Genet* 2007;39(9):1065-7.