

Genetics of Complex Diseases: Rheumatic Diseases

2008

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I. GENETICS OF COMPLEX DISEASE:

A. Concepts in genetic epidemiology: large scale association and genome wide association

1. Burton PR, Tobin MD, Hopper JL. Key concepts in genetic epidemiology. Lancet 2005;366:941-51. Review. Erratum in: Lancet 2006;367:28. [PUB MED LINK](#)
2. Pearson TA, Manolio TA. How to interpret a genome-wide association study. JAMA 2008;299:1335-44. Erratum in: JAMA 2008;299:2150. [PUB MED LINK](#)
3. Kraft P, Cox DG. Study designs for genome-wide association studies. Adv Genet 2008;60:465-504. [PUB MED LINK](#)
4. Ehm MG, Nelson MR, Spurr NK. Guidelines for conducting and reporting whole genome/large-scale association studies. Hum Mol Genet 2005;14:2485-8. [PUB MED LINK](#)

B. Utility of low risk alleles

1. Offit K. Genomic profiles for disease risk: predictive or premature? JAMA 2008;299:1353-5. [PUB MED LINK](#)

C. Internet genetic resources

1. Uhlmann WR, Guttmacher AE. Key Internet genetics resources for the clinician. JAMA 2008;299:1356-8. [PUB MED LINK](#)

II. SERONEGATIVE SPONDYLOARTHROPATHIES

A. Landmark Paper

1. Brewerton DA, Hart FD, Nicholls A, Caffrey M, James DC, Sturrock RD. Ankylosing spondylitis and HL-A 27. Lancet 1973;1:904-7. [PUB MED LINK](#)

B. Reviews

1. Brown MA. Breakthroughs in genetic studies of ankylosing spondylitis. Rheumatology (Oxford) 2008;47:132-7. [PUB MED LINK](#)

2. Brionez TF, Reveille JD. The contribution of genes outside the major histocompatibility complex to susceptibility to ankylosing spondylitis. *Curr Opin Rheumatol* 2008;20:384-91. [PUB MED LINK](#)

C. Genetic studies - Meta-analysis of genome wide linkage studies

1. Carter KW, Pluzhnikov A, Timms AE, Miceli-Richard C, Bourgain C, Wordsworth BP, et al. Combined analysis of three whole genome linkage scans for Ankylosing Spondylitis. *Rheumatology (Oxford)* 2007;46:763-71. [PUB MED LINK](#)

D. Genome wide association in AS

1. Wellcome Trust Case Control Consortium; Australo-Anglo-American Spondylitis Consortium (TASC), Burton PR, Clayton DG, Cardon LR, Craddock N, et al. Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. *Nat Genet* 2007;39:1329-37. [PUB MED LINK](#)

E. Genetics of extra-articular features

1. Duffin KC, Chandran V, Gladman DD, Krueger GG, Elder JT, Rahman P. Genetics of Psoriasis and Psoriatic Arthritis: Update and Future Direction. *J Rheumatol* 2008;35:1449-1453. [PUB MED LINK](#)
2. Liu Y, Helms C, Liao W, Zaba LC, Duan S, Gardner J, et al. A genome-wide association study of psoriasis and psoriatic arthritis identifies new disease loci. *PLoS Genet* 2008;4:e1000041. [PUB MED LINK](#)
3. Cho JH. The genetics and immunopathogenesis of inflammatory bowel disease. *Nat Rev Immunol.* 2008;8:458-66. [PUB MED LINK](#)

III. RHEUMATOID ARTHRITIS

A. Landmark Paper

1. Stastny P. Association of the B-cell alloantigen DRw4 with rheumatoid arthritis. *N Engl J Med* 1978;298:869-71. [PUB MED LINK](#)
2. Gregersen PK, Silver J, Winchester RJ. The shared epitope hypothesis. An approach to understanding the molecular genetics of susceptibility to rheumatoid arthritis. *Arthritis Rheum* 1987;30:1205-13. [PUB MED LINK](#)
3. Begovich AB, Carlton VE, Honigberg LA, Schrodi SJ, Chokkalingam AP, Alexander HC, et al. A missense single-nucleotide polymorphism in a gene encoding a protein tyrosine phosphatase (PTPN22) is associated with rheumatoid arthritis. *Am J Hum Genet* 2004;75:330-7. [PUB MED LINK](#)

B. Reviews

1. Gregersen PK. T-cell receptor-major histocompatibility complex genetic interactions in rheumatoid arthritis. *Rheum Dis Clin North Am* 1992;18:793-807. [PUB MED LINK](#)

2. Bowes J, Barton A. Recent advances in the genetics of RA susceptibility. *Rheumatology (Oxford)* 2008;47:399-402. [PUB MED LINK](#)

C. Genome-wide Linkage studies

1. Jawaheer D, Seldin MF, Amos CI, Chen WV, Shigeta R, Etzel C, et al. Screening the genome for rheumatoid arthritis susceptibility genes: a replication study and combined analysis of 512 multicase families. *Arthritis Rheum* 2003;48:906-16. [PUB MED LINK](#)
2. Etzel CJ, Chen WV, Shepard N, Jawaheer D, Cornelis F, Seldin MF, et al. Genome-wide meta-analysis for rheumatoid arthritis. *Hum Genet* 2006;119:634-41. [PUB MED LINK](#)

D. Genome-Wide Association studies

1. Wellcome Trust Case Control Consortium. Genome-wide association study of 14,000 cases of seven common diseases and 3,000 shared controls. *Nature* 2007;447:661-78. [PUB MED LINK](#)
2. Dieguez-Gonzalez R, Calaza M, Perez-Pampin E, de la Serna AR, Fernandez-Gutierrez B, Castañeda S, et al. Association of interferon regulatory factor 5 haplotypes, similar to that found in systemic lupus erythematosus, in a large subgroup of patients with rheumatoid arthritis. *Arthritis Rheum* 2008;58:1264-74. [PUB MED LINK](#)
3. Plenge RM, Cotsapas C, Davies L, Price AL, de Bakker PI, Maller J, et al. Two independent alleles at 6q23 associated with risk of rheumatoid arthritis. *Nat Genet* 2007;39:1477-82. [PUB MED LINK](#)
4. Plenge RM, Seielstad M, Padyukov L, Lee AT, Remmers EF, Ding B, et al. TRAF1-C5 as a risk locus for rheumatoid arthritis--a genomewide study. *N Engl J Med* 2007;357:1199-209. [PUB MED LINK](#)

E. Gene-Gene and Gene-Environment Interaction

1. Kallberg H, Padyukov L, Plenge RM, Ronnelid J, Gregersen PK, van der Helm-van Mil AH, et al. Gene-gene and gene-environment interactions involving HLA-DRB1, PTPN22, and smoking in two subsets of rheumatoid arthritis. *Am J Hum Genet* 2007;80:867-75. [PUB MED LINK](#)

IV. SYSTEMIC LUPUS ERYTHEMATOSUS

A. Reviews

1. Sestak AL, Nath SK, Sawalha AH, Harley JB. Current status of lupus genetics. *Arthritis Res Ther* 2007;9:210. [PUB MED LINK](#)
2. Gregersen PK, Behrens TW. Genetics of autoimmune diseases--disorders of immune homeostasis. *Nat Rev Genet* 2006;7:917-28. [PUB MED LINK](#)

B. Genome-wide linkage studies

1. Forabosco P, Gorman JD, Cleveland C, Kelly JA, Fisher SA, Ortmann WA, et al. Meta-analysis of genome-wide linkage studies of systemic lupus erythematosus. *Genes Immun* 2006;7:609-14. [PUB MED LINK](#)
2. Lee YH, Nath SK. Systemic lupus erythematosus susceptibility loci defined by genome scan meta-analysis. *Hum Genet* 2005;118:434-43. [PUB MED LINK](#)

C. Genome-wide Association studies

1. International Consortium for Systemic Lupus Erythematosus Genetics (SLEGEN), Harley JB, Alarcón-Riquelme ME, Criswell LA, Jacob CO, Kimberly RP, et al. Genome-wide association scan in women with systemic lupus Erythematosus identifies susceptibility variants in ITGAM, PXX, KIAA1542 and other loci. *Nat Genet* 2008;40:204-10. [PUB MED LINK](#)
2. Hom G, Graham RR, Modrek B, Taylor KE, Ortmann W, Garnier S, et al. Association of systemic lupus erythematosus with C8orf13-BLK and ITGAM-ITGAX. *N Engl J Med* 2008;358:900-9. [PUB MED LINK](#)
3. Kozyrev SV, Abelson AK, Wojcik J, Zaghlool A, Linga Reddy MV, Sanchez E, et al. Functional variants in the B-cell gene BANK1 are associated with systemic lupus erythematosus. *Nat Genet* 2008;40:211-6. Erratum in: *Nat Genet* 2008;40:484. Barizzzone, Nadia [corrected to Barizzzone, Nadia]. [PUB MED LINK](#)

D. Other

1. Kyogoku C, Langefeld CD, Ortmann WA, Lee A, Selby S, Carlton VE, et al. Genetic association of the R620W polymorphism of protein tyrosine phosphatase PTPN22 with human SLE. *Am J Hum Genet* 2004;75:504-7. [PUB MED LINK](#)
2. Graham RR, Kozyrev SV, Baechler EC, Reddy MV, Plenge RM, Bauer JW, et al. A common haplotype of interferon regulatory factor 5 (IRF5) regulates splicing and expression and is associated with increased risk of systemic lupus erythematosus. *Nat Genet* 2006;38:550-5. [PUB MED LINK](#)
3. Graham RR, Kyogoku C, Sigurdsson S, Vlasova IA, Davies LR, Baechler EC, et al. Three functional variants of IFN regulatory factor 5 (IRF5) define risk and protective haplotypes for human lupus. *Proc Natl Acad Sci U S A* 2007;104:6758-63. [PUB MED LINK](#)
4. Remmers EF, Plenge RM, Lee AT, Graham RR, Hom G, Behrens TW, et al. STAT4 and the risk of rheumatoid arthritis and systemic lupus erythematosus. *N Engl J Med* 2007;357:977-86. [PUB MED LINK](#)
5. Crow MK, Kirou KA, Wohlgemuth J. Microarray analysis of interferon-regulated genes in SLE. *Autoimmunity* 2003;36:481-90. [PUB MED LINK](#)

V. SYSTEMIC SCLEROSIS

1. Agarwal SK, Tan FK, Arnett FC. Genetics and genomic studies in scleroderma (systemic sclerosis). *Rheum Dis Clin North Am* 2008;34:17-40; v. [PUB MED LINK](#)
2. Fonseca C, Lindahl GE, Ponticos M, Sestini P, Renzoni EA, Holmes AM, et al. A polymorphism in the CTGF promoter region associated with systemic sclerosis. *N Engl J Med* 2007;357:1210-20. [PUB MED LINK](#)

VI. OSTEOARTHRITIS

A. Reviews

1. Valdes AM, Spector TD. The contribution of genes to osteoarthritis. *Rheum Dis Clin North Am* 2008;34:581-603. [PUB MED LINK](#)
2. Ikegawa S. New gene associations in osteoarthritis: what do they provide, and where are we going? *Curr Opin Rheumatol* 2007;19:429-34. [PUB MED LINK](#)

B. Genome Wide association studies

1. Valdes AM, Loughlin J, Timms KM, van Meurs JJ, Southam L, Wilson SG, et al. Genome-wide association scan identifies a prostaglandin-endoperoxide synthase 2 variant involved in risk of knee osteoarthritis. *Am J Hum Genet* 2008;82:1231-40. [PUB MED LINK](#)
2. Spector TD, Reneland RH, Mah S, Valdes AM, Hart DJ, Kammerer S, et al. Association between a variation in LRCH1 and knee osteoarthritis: a genome-wide single-nucleotide polymorphism association study using DNA pooling. *Arthritis Rheum* 2006;54:524-32. [PUB MED LINK](#)

VII. OSTEOPOROSIS

A. Reviews

1. Williams FM, Spector TD. Recent advances in the genetics of osteoporosis. *J Musculoskelet Neuronal Interact* 2006;6:27-35. [PUB MED LINK](#)

B. Genome Wide Association studies

2. Richards JB, Rivadeneira F, Inouye M, Pastinen TM, Soranzo N, Wilson SG, et al. Bone mineral density, osteoporosis, and osteoporotic fractures: a genome-wide association study. *Lancet* 2008;371:1505-12. [PUB MED LINK](#)
3. Styrkarsdottir U, Halldorsson BV, Gretarsdottir S, Gudbjartsson DF, Walters GB, Ingvarsson T, et al. Multiple genetic loci for bone mineral density and fractures. *N Engl J Med* 2008;358:2355-65. [PUB MED LINK](#)

VIII. PHARMACOGENETICS

1. Nebert DW, Zhang G, Vesell ES. From human genetics and genomics to pharmacogenetics and pharmacogenomics: past lessons, future directions. *Drug Metab Rev* 2008;40:187-224. [PUB MED LINK](#)
2. Ranganathan P. Pharmacogenomics in rheumatoid arthritis. *Methods Mol Biol.* 2008;448:413-35. [PUB MED LINK](#)
3. Sahasranaman S, Howard D, Roy S. Clinical pharmacology and pharmacogenetics of thiopurines. *Eur J Clin Pharmacol* 2008;64:753-67. [PUB MED LINK](#)
4. Ranganathan P, McLeod HL. Methotrexate pharmacogenetics: the first step toward individualized therapy in rheumatoid arthritis. *Arthritis Rheum* 2006;54:1366-77. [PUB MED LINK](#)
5. Plenge RM, Criswell LA. Genetic variants that predict response to anti-tumor necrosis factor therapy in rheumatoid arthritis: current challenges and future directions. *Curr Opin Rheumatol* 2008;20:145-52. [PUB MED LINK](#)