

Literature Cited

- Abecasis GR, Cherny SS, Cookson WO, Cardon LR (2002) Merlin – rapid analysis of dense genetic maps using sparse gene flow trees. *Nature Genetics* 30:97–101
- Abreu PC, Greenberg DA, Hodge SE (1999) Direct power comparisons between simple LOD scores and NPL scores for linkage analyses in complex diseases. *American Journal of Human Genetics* 65:847–857
- AGP (2007) Mapping autism risk loci using genetic linkage and chromosomal rearrangements. *Nature Genetics* (in press)
- Akaike H (1974) A new look at the statistical model identification. *IEEE Transactions on automatic control* 19:716–723
- Almasy L, Blangero J (1998) Multipoint quantitative-trait linkage analysis in general pedigrees. *American Journal of Human Genetics* 62:1198–1211
- Amos CI (1994) Robust variance-components approach for assessing genetic linkage in pedigrees. *American Journal of Human Genetics* 54:535–543
- Amos CI, Schete S, Chen J, Yu RK (2003) Positional identification of microdeletions with genetic markers. *Human Heredity* 56:107–118
- Ardlie KG, Kruglyak L, Seielstad M (2002) Patterns of linkage disequilibrium in the human genome. *Nature Reviews Genetics* 3:299–309
- Atwood LD, Heard-Costa NL (2003) Limits of fine-mapping a quantitative trait. *Genetic Epidemiology* 24:99–106
- Badzioch MD, Goode EL, Jarvik GP (2005) The role of parametric linkage methods in complex trait analyses using microsatellites. *Biomed Central Genetics* 30 (Suppl 1):S48
- Basu S, Di Y, Thompson EA (2007) Tests for linkage detection in pedigrees. Submitted
- Basu S, Wijsman EM, Thompson EA (2002) Allele-sharing methods on large pedigrees. *Genetic Epidemiology* 23:267
- Baum LE, Petrie T, Soules G, Weiss N (1970) A maximization technique occurring in the statistical analysis of probabilistic functions on Markov chains. *Annals of Mathematical Statistics* 41:164–171
- Biernacka JM, Sun L, Bull SB (2005) Simultaneous localization of two linked disease-susceptibility genes. *Genetic Epidemiology* 28:33–47
- Biswas S, Papachristou C, Irwin ME, Lin S (2003) Linkage analysis of the simulated data: Evaluations and comparisons of methods. *Biomed Central Genetics* 4 (Suppl 1):S70
- Blacker D, Bertram L, Saunders AJ, Moscarillo TJ, Albert MS, Wiener H, Perry RT, Collins JS, Harrell LE, Go RCP, et al (2003) Results of a high-resolution genome screen of 437 Alzheimer’s Disease families. *Human Molecular Genetics* 12:23–32
- Blangero J, Williams JT, Almasy L (2000) Robust LOD scores for variance component-based linkage analysis. *Genetic Epidemiology* 19:S8–S14. Suppl. 1
- Boehnke M (1994) Limits of resolution of genetic linkage studies: Implications for the positional cloning of human disease genes. *Am J Hum Genet* 55:379–390
- Boehnke M, Cox NJ (1997) Accurate inference of relationships in sib-pair linkage studies. *American Journal of Human Genetics* 61:423–429
- Boerwinkle E, Brown CA, Carrejo M, Ferrell R, Hanis C, Hutchinson R, Kardia S, Sing C, Turner S, Weder A, et al (2002) Multi-center genetic study of hypertension - The Family Blood Pressure Program (FBPP). *Hypertension* 39:3–9

- Broman KW, Murray JC, Sheffield VC, White RL, Weber JL (1998) Comprehensive human genetic maps: Individual and sex-specific variation in recombination. *American Journal of Human Genetics* 63:861–869
- Brown MS, Goldstein JL (1986) A receptor-mediated pathway for cholesterol homeostasis. *Science* 232:34–47
- Browning SG (1998) Relationship information contained in gamete identity by descent data. *Journal of Computational Biology* 5:323–334
- Chang YPC, Kim JDO, Schwander K, Rao DC, Miller MB, Weder AB, Cooper RS, Schork NJ, Province MA, Morrison AC, et al (2006) The impact of data quality on the identification of complex disease genes: experience from the Family Blood Pressure Program. *European Journal of Human Genetics* 14:469–477
- Chapman NH, Leutenegger A, Badzioch MD, Bogdan M, Conlon EM, Daw EW, Gagnon F, Li N, Maia JM, Wijsman EM, Thompson EA (2001) The importance of connections: Joining components of the Hutterite pedigree. *Genetic Epidemiology* 21 (Suppl 1):S230–S235
- Chapman NH, Wijsman EM (1998) Genome screens using linkage disequilibrium tests: Optimal marker characteristics and feasibility. *American Journal of Human Genetics* 63:1872–1885
- Churchill GA, Doerge RW (1994) Empirical threshold values for quantitative trait mapping. *Genetics* 138:963–971
- Cohen JC, Kiss RS, Pertsemlidis A, Marcel YL, McPherson R, Hobbs HH (2004) Multiple rare alleles contribute to low plasma levels of HDL cholesterol. *Science* 305:869–872
- Conrad DF, Andrews TD, Carter NP, Hurles ME, Pritchard JK (2006) A high-resolution survey of deletion polymorphism in the human genome. *Nature Genetics* 38:75–81
- Corder EH, Saunders AM, Strittmatter WJ, Schmechel DE, Gaskell PC, Small GW, Roses AD, Haines JL, Pericak-Vance MA (1993) Gene dose of Apolipoprotein-E type-4 allele and the risk of Alzheimer’s Disease in late onset families. *Science* 261:921–923
- Davis S, Schroeder M, Goldin LR, Weeks DE (1996) Nonparametric simulation-based statistics for detecting linkage in general pedigrees. *American Journal of Human Genetics* 58:867–880
- Daw EW, Thompson EA, Wijsman EM (2000) Bias in multipoint linkage analysis arising from map misspecification. *Genetic Epidemiology* 19:366–380
- (2003) A score for Bayesian genome screening. *Genetic Epidemiology* 24:181–190
- Dempster AP, Laird NM, Rubin DB (1977) Maximum likelihood from incomplete data via the EM algorithm (with Discussion). *Journal of the Royal Statistical Society B* 39:1–37
- Dewan A, Liu MG, Hartman S, Zhang SSM, Liu DTL, Zhao C, Tam POS, Chan WM, Lam DSC, Snyder M, Barnstable C, Pang CP, Hoh J (2006) HTRA1 promoter polymorphism in wet age-related macular degeneration. *Science* 314:989–992
- Diao G, Lin DY (2006) Semiparametric variance-component models for linkage and association analyses of censored trait data. *Genetic Epidemiology* 30:570–581
- Dietter J, Mattheisen M, Furst R, Ruschendorf F, Wienker TF, Strauch K (2007) Linkage analysis using sex-specific recombination fractions with GENEHUNTER-MODSCORE. *Bioinformatics* 23:64–70
- Dietter J, Spiegel A, Mey DA, Pflug HJ, Al-Kateb H, Hoffmann K, Wienker TF, Strauch K (2004) Efficient two-trait-locus linkage analysis through program optimization and parallelization: application to hypercholesterolemia. *European Journal of Human Genetics* 12:542–550
- Donis-Keller H, Green P, Helms C, Cartinhour S, Weiffenbach B, Stephens K, Keith TP, Bowden BW, Smith DR, Lander ES, Botstein D, et al (1987) A genetic linkage map of the human genome. *Cell* 51:319–337
- Edwards AO, Ritter R, Abel KJ, Manning A, Panhuysen C, Farrer LA (2005) Complement factor H polymorphism and age-related macular degeneration. *Science* 308:421–424

- Edwards D, Havranek T (1987) A fast model selection procedure for large families of models. *Journal of the American Statistical Association* 82:205–213
- Elbaz A, Nelson LM, Payami H, Ioannidis JPA, Fiske BK, Annesi G, Belin AC, Factor SA, Ferrarese C, Hadjigeorgiou GM, et al (2006) Lack of replication of thirteen single-nucleotide polymorphisms implicated in Parkinson’s disease: a large-scale international study. *Lancet Neurology* 5:917–923
- Elston RC, Stewart J (1971) A general model for the analysis of pedigree data. *Human Heredity* 21:523–542
- Emahazion T, Feuk L, Jobs M, Sawyer SL, Fredman D, St Clair D, Prince J, Brookes A (2001) SNP association studies in Alzheimer’s disease highlight problems for complex disease analysis. *Trends in Genetics* 17:407–413
- Epstein MP, Lin XH, Boehnke M (2003) A tobit variance-component method for linkage analysis of censored trait data. *American Journal of Human Genetics* 72:611–620
- Falk CT (2001) Introduction: Haplotype analysis of simulated genetic analysis workshop 12 data. *Genetic Epidemiology* 21 (Suppl 1):S552–S553
- Feuk L, Marshall CR, Wintle RF, Scherer SW (2006) Structural variants: changing the landscape of chromosomes and design of disease studies. *Human Molecular Genetics* 15:R57–R66. Sp. Iss. 1
- Field LL (2002) Genetic linkage and association studies of Type I diabetes: Challenges and rewards. *Diabetologia* 45:21–35
- Fingerlin TE, Abecasis GR, Boehnke M (2006) Using sex-averaged genetic maps in multipoint linkage analysis when identity-by-descent status is incompletely known. *Genetic Epidemiology* 30:384–396
- Fishelson M, Geiger D (2004) Optimizing exact linkage computations. *Journal of Computational Biology* 11:263–275
- Fisher RA (1934) The amount of information supplied by records of families as a function of the linkage in the population sampled. *Annals of Eugenics* 6:66–70
- Fisher SA, Abecasis GR, Yashar BM, Zarepari S, Swaroop A, Iyengar SK, Klein BEK, Klein R, Lee KE, Majewski J, et al (2005) Meta-analysis of genome scans of age-related macular degeneration. *Human Molecular Genetics* 14:2257–2264
- Forrest, Feingold (2000) Composite statistics for QTL mapping with moderately discordant sibling pairs. *American Journal of Human Genetics* 66:2020–2020
- Fridman E, Carrari F, Liu YS, Fernie AR, Zamir D (2004) Zooming in on a quantitative trait for tomato yield using interspecific introgressions. *Science* 305:1786–1789
- Gabriel SB, Salomon R, Pelet A, Angrist M, Amiel J, Fornage M, Attie-Bitach T, Olson JM, Hofstra R, Buys C, Steffann J, Munnich A, Lyonnet S, Chakravarti A (2002) Segregation at three loci explains familial and population risk in Hirschsprung disease. *Nature Genetics* 31:89–93
- George AW, Wijsman EM, Thompson EA (2005) MCMC multilocus lod scores: Application of a new approach. *Human Heredity* 59:98–108
- Geyer CJ, Meeden GD (2005) Fuzzy and randomized confidence intervals and p -values (with Discussion). *Statistical Science* 20:358–87
- Giglio S, Broman KW, Matsumoto N, Calvari V, Gimelli G, Neumann T, Ohashi H, Voullaire L, Larizza D, Giorda R, et al (2001) Olfactory receptor-gene clusters, genomic-inversion polymorphisms, and common chromosome rearrangements. *American Journal of Human Genetics* 68:874–883
- Glazier AM, Nadeau JH, Aitman T (2002) Finding genes that underlie complex traits. *Science* 298:2345–2349
- Goate A, Harlin CMC, Mullan M, Brown J, Crawford F, Fidani L, Giuffra L, Haynes A, Irving N, James L, et al (1991) Segregation of a missense mutation in the amyloid precursor protein gene with familial Alzheimer’s disease. *Nature* 349:704–706

- Goldgar DE, Easton DF (1997) Optimal strategies for mapping complex diseases in the presence of multiple loci. *American Journal of Human Genetics* 60:1222–1232
- Goldin LR (2001) Introduction: Linkage analysis of quantitative traits. *Genetic Epidemiology* 21 (Suppl 1):S459–S460
- Gretarsdottir S, Sveinbjornsdottir S, Jonsson HH, Jakobsson F, Einarsdottir E, Agnarsson U, Shkolny D, Einarsson G, Gudjonsdottir HM, Valdimarsson EM, et al (2002) Localization of a susceptibility gene for common forms of stroke to 5q12. *American Journal of Human Genetics* 70:593–603
- Guo SW, Thompson EA (1994) Monte Carlo estimation of mixed models for large complex pedigrees. *Biometrics* 50:417–432
- Haines JL, Hauser MA, Schmidt S, Scott WK, Olson LM, Gallins P, Spencer KL, Kwan SY, Nouredine M, Gilbert JR, Schmetz-Boutaud N, Agarwal A, Postel EA, Pericak-Vance MA (2005) Complement factor H variant increases the risk of age-related macular degeneration. *Science* 308:419–421
- Haldane JBS (1934) Methods for the detection of autosomal linkage in man. *Annals of Eugenics* 6:26–65
- Haldane JBS, Smith CAB (1947) A new estimate of the linkage between the genes for clour-blindness and haemophilia in man. *Annals of Eugenics* 14:10–31
- Haseman JK, Elston RC (1972) The investigation of linkage between a quantitative trait and a marker locus. *Behavior Genetics* 2:3–19
- Heath SC (1997) Markov chain Monte Carlo segregation and linkage analysis for oligogenic models. *American Journal of Human Genetics* 61:748–760
- Hirschhorn JN, Lohmueller K, Byrne E, Hirschhorn K (2002) A comprehensive review of genetic association studies. *Genetics in Medicine* 4:45–61
- Huang JZ, Liu N, Pourahmadi M, Liu L (2006) Covariance matrix selection and estimation via penalised Normal likelihood. *Biometrika* 93:85–98
- Igo RP, Chapman NH, Berninger VW, Matsushita M, Brkanac Z, Rothstein JH, Holzman T, Nielsen K, Raskind W, Wijsman EM (2006) Genomewide scan for real-word reading subphenotypes of dyslexia: Novel chromosome 13 locus and genetic complexity. *American Journal of Medical Genetics Neuropsychiatric Genetics* 141B:15–27
- International Hapmap Consortium (2005) A haplotype map of the human genome. *Nature* 237:1299–1319
- Jansen RC (2001) Quantitative trait loci in inbred lines. In DJ Balding, M Bishop, C Cannings, eds., *Handbook of Statistical Genetics*, 567–597. Wiley, London
- Jung J, Weeks DE, Feingold E (2006) Gene-dropping vs. empirical variance estimation for allele-sharing linkage statistics. *Genetic Epidemiology* 30:652–665
- Klein ML, Schultz DW, Edwards A, Matise TC, Rust K, Berselli CB, Trzuppek K, Weleber RG, Ott J, Wirtz MK, Acott TS (1998) Age-related macular degeneration - Clinical features in a large family and linkage to chromosome 1q. *Archives of Ophthalmology* 116:1082–1088
- Klein RJ, Zeiss C, Chew EY, Tsai JY, Sackler RS, Haynes C, Henning AK, SanGiovanni JP, Mane SM, Mayne ST, Bracken MB, Ferris FL, Ott J, Barnstable C, Hoh J (2005) Complement factor H polymorphism in age-related macular degeneration. *Science* 308:385–389
- Kong A, Cox NJ (1997) Allele-sharing models: Lod scores and accurate linkage tests. *American Journal of Human Genetics* 61:1179–1188
- Kreiner S (1987) Analysis of multidimensional contingency tables by exact conditional tests: techniques and strategies. *Scandinavian Journal of Statistics theory and applications* 14:97–112
- Kruglyak L, Daly MJ, Reeve-Daly MP, Lander ES (1996) Parametric and nonparametric linkage analysis: A unified multipoint approach. *American Journal of Human Genetics* 58:1347–1363

- Kruglyak L, Lander ES (1995) Complete multipoint sib-pair analysis of qualitative and quantitative traits. *American Journal of Human Genetics* 57:439–454
- Lander E, Kruglyak L (1995) Genetic dissection of complex traits: guidelines for interpreting and reporting linkage results. *Nature Genetics* 11:241–247
- Lander ES, Green P (1987) Construction of multilocus genetic linkage maps in humans. *Proceedings of the National Academy of Sciences USA* 84:2363–2367
- Lange K, Sobel E (1991) A random walk method for computing genetic location scores. *American Journal of Human Genetics* 49:1320–1334
- Lathrop GM, Lalouel JM, Julier C, Ott J (1984) Strategies for multilocus linkage analysis in humans. *Proceedings of the National Academy of Sciences USA* 81:3443–3446
- Lauritzen SL (1996) *Graphical Models*. Oxford University Press, Oxford UK
- Levy-Lahad E, Wasco W, Poorkaj P, Romano DM, Oshima J, Pettingell WH, Yu CE, Jondro PD, Schmidt SD, Wang K, et al (1995a) Candidate gene for the chromosome 1 familial Alzheimer’s disease locus. *Science* 269:973–977
- Levy-Lahad E, Wijsman EM, Nemens E, Anderson L, Goddard KA, Weber JL, Bird TD, Schellenberg GD (1995b) A familial Alzheimer’s disease locus on chromosome 1. *Science* 269:970–973
- Li MY, Boehnke M, Abecasis GR, Song PXX (2006) Quantitative trait linkage analysis using Gaussian copulas. *Genetics* 173:2317–2327
- Logue MW, Vieland VJ (2004) A new method for computing the multipoint posterior probability of linkage. *Human Heredity* 57:90–99
- Longmate JA (2001) Complexity and power in case-control association studies. *American Journal of Human Genetics* 68:1229–1237
- Lorincz MT (2006) Clinical implications of Parkinson’s disease genetics. *Seminars in Neurology* 26:492–498
- Mackay TFC (2001) Quantitative trait loci in *Drosophila*. *Nature Reviews Genetics* 2:11–20
- Madigan D, York J, Allard D (1995) Bayesian graphical models for discrete data. *International Statistical Review* 63:215–232
- Majewski J, Schultz DW, Weleber RG, Schain MB, Edwards AO, Matise TC, Acott TS, Ott J, Klein ML (2003) Age-related macular degeneration - a genome scan in extended families. *American Journal of Human Genetics* 73:540–550
- McPeck MS (1999) Optimal allele-sharing statistics for genetic mapping using affected relatives. *Genetic Epidemiology* 16:225–249
- Meinshausen N, Bühlmann P (2006) High-dimensional graphs and variable selection with the LASSO. *Annals of Statistics* 34:1436–1462
- Morris RW, Kaplan NL (2002) On the advantage of haplotype analysis in the presence of multiple disease susceptibility alleles. *Genetic Epidemiology* 23:221–233
- Morton NE (1955) Sequential tests for the detection of linkage. *American Journal of Human Genetics* 7:277–318
- Nicolae DL, Kong A (2004) Measuring the relative information in allele-sharing linkage studies. *Biometrics* 60:368–375
- O’Brien SJ, Nelson GW, Winkler CA, Smith MW (2000) Polygenic and multifactorial disease gene association in man: Lessons from AIDS. *Annual Review of Genetics* 34:563–591
- O’Connell JR, Weeks DE (1995) The algorithm for rapid exact multilocus linkage analysis via genotype set-recoding and fuzzy inheritance. *Nature Genetics* 11:402–408
- Ott J (1974) Estimation of the recombination frequency in human pedigrees: Efficient computation of the likelihood for human linkage studies. *American Journal of Human Genetics* 26:588–597

- (1999) *Analysis of Human Genetic Linkage*. 3 rd. ed., The Johns Hopkins University Press, Baltimore, MD
- Owen MJ, Williams NM, O'Donovan MC (2004) The molecular genetics of schizophrenia: findings promise new insights. *Molecular Psychiatry* 9:14–27
- Penrose LS (1935) The detection of autosomal linkage in data which consist of pairs of brothers and sisters of unspecified parentage. *Annals of Eugenics* 6:133–138
- Polymeropoulos MH, Lavedan C, Leroy E, Ide SE, Dehejia A, Dutra A, Pike B, Root H, Rubenstein J, Boyer R, et al (1997) Mutation in the alpha-synuclein gene identified in families with Parkinson's disease. *Science* 276:2045–2047
- Province MA, Rice TK, Borecki IB, Gu C, Kraja A, Rao DC (2003) Multivariate and multilocus variance components method, based on structural relationships to assess quantitative trait linkage via SEGPATH. *Genetic Epidemiology* 24:128–138.
- Rao DC, Keats BJB, Morton NE, Yee S, Lew R (1978) Variability of human linkage data. *American Journal of Human Genetics* 30:516–529
- Redon R, Ishikawa S, Fitch KR, Feuk L, Perry GH, Andrews TD, Fiegler H, Shapero MH, Carson AR, Chen WW, et al (2006) Global variation in copy number in the human genome. *Nature* 444:444–454
- Risch N (1990) Linkage strategies for genetically complex traits. III. The effect of marker polymorphism on analysis of affected relative pairs. *American Journal of Human Genetics* 46:242–253
- Risch N, Merikangas K (1996) The future of genetic studies of complex human diseases. *Science* 273:1516–1517
- Rivera A, Fisher SA, Fritsche LG, Keilhauer CN, Lichtner P, Meitinger T, Weber BHF (2005) Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. *Human Molecular Genetics* 14:3227–3236
- Rogaeva E (2002) The solved and unsolved mysteries of the genetics of early-onset Alzheimer's disease. *Neuro-molecular Medicine* 2:1–10
- Rovelet-Lecruz A, Hannequin D, Raux G, Le Meur N, Laquerriere A, Vital A, Dumanchin C, Feuillette S, Brice A, Vercelletto M, Dubas F, Frebourg T, Campion D (2006) APP locus duplication causes autosomal dominant early-onset Alzheimer disease with cerebral amyloid angiopathy. *Nature Genetics* 38:24–26
- Scheet P, Stephens M (2006) A fast and flexible statistical model for large-scale population genotype data: Applications to inferring missing genotypes and haplotypic phase. *American Journal of Human Genetics* 78:629–644
- Schellenberg GD, Dawson G, Sung YJ, Estes A, Munson J, Rosenthal E, Rothstein J, Flodman P, Smith M, Coon H, et al (2006) Evidence for multiple loci from a genome scan of autism kindreds. *Molecular Psychiatry* 11:1049–1060
- Schork NJ, Boehnke M, Terwilliger JD, Ott J (1993) Two-trait-locus linkage analysis: a powerful strategy for mapping complex genetic traits. *American Journal of Human Genetics* 53:1127–36
- Schwarz G (1978) Estimating the Dimension of a Model. *The Annals of Statistics* 6:461–464
- Seddon JM, Santangelo SL, Book K, Chong S, Cote J (2003) A genomewide scan for age-related macular degeneration provides evidence for linkage to several chromosomal regions. *American Journal of Human Genetics* 73:780–790
- Sham PC, Lin MW, Zhao JH, Curtis D (2000) Power comparison of parametric and nonparametric linkage tests in small pedigrees. *American Journal of Human Genetics* 66:1661–1668
- Sharp AJ, Cheng Z, Eichler EE (2006) Structural variation of the human genome. *Annual Review of Genomics and Human Genetics* 7:407–442
- Sherrington R, Rogaev EI, Liang Y, Rogaeva EA, Levesque G, Ikeda M, Chi H, Lin C, Li G, Holman K, et al (1995) Cloning of a gene bearing missense mutations in early-onset familial Alzheimer's disease. *Nature* 375:754–760

- Sieberts SK, Wijsman EM, Thompson EA (2002) Relationship inference from trios of individuals in the presence of typing error. *American Journal of Human Genetics* 70:170–180
- Sieh W, Basu S, Fu AQ, Rothstein JH, Scheet PA, Stewart W, Sung YJ, Thompson EA, Wijsman EM (2005) Comparison of marker types and map assumptions using MCMC-based linkage analysis of COGA data. *Biomed Central Genetics* 6:S11
- Sieh W, Yu CE, Bird TD, Schellenberg GD, Wijsman EM (2007) Accounting for linkage disequilibrium among markers in linkage analysis: Impact of haplotype frequency estimation and molecular haplotypes for a gene in a candidate region for Alzheimer’s Disease. *Human Heredity* 63:26–34
- Skipper L, Farrer M (2002) Parkinson’s genetics: Molecular insights for the new millennium. *Neurotoxicology* 23:503–514
- Smith CAB (1953) Detection of linkage in human genetics. *Journal of the Royal Statistical Society B* 15:153–192
- Smyth DJ, Cooper JD, Bailey R, Field S, Burren O, Smink LJ, Guja C, Ionescu-Tirgoviste C, Widmer B, Dunger DB, et al (2006) A genome-wide association study of nonsynonymous SNPs identifies a type 1 diabetes locus in the interferon-induced helicase (IFIH1) region. *Nature Genetics* 38:617–619
- Sobel E, Sengal H, Weeks DE (2001) Multipoint estimation of identity-by-descent probabilities at arbitrary positions among marker loci on general pedigrees. *Human Heredity* 52:121–131
- St. George-Hyslop PH, Tanzi RE, Polinsky RJ, Haines JL, Nee L, Watkins PC, Myers RH, Feldman RG, Pollen D, Drachman D, et al (1987) The genetic defect causing familial Alzheimer’s disease maps on chromosome 21. *Science* 235:885–890
- Stefansson H, Helgason A, Thorleifsson G, Steinthorsdottir V, Masson G, Barnard J, Baker A, Jonasdottir A, Ingason A, Gudnadottir VG, et al (2005) A common inversion under selection in Europeans. *Nature Genetics* 37:129–137
- Stewart WCL, Thompson EA (2006) Improving estimates of genetic maps: A maximum likelihood approach. *Biometrics* 62:728–734
- Suarez BK, Rice J, Reich T (1978) The generalized sib pair IBD distribution: Its use in the detection of linkage. *Annals of Human Genetics* 42:87–94
- Sung YJ, Thompson EA, Wijsman E (2007) MCMC-based linkage analysis for complex traits on general pedigrees: Multipoint analysis with a two-locus model and a polygenic component. *Genetic Epidemiology* 31:103–114
- Tanzi RE, Bertram L (2005) Twenty years of the Alzheimer’s disease amyloid hypothesis: A genetic perspective. *Cell* 120:545–555
- Terwilliger JD, Weiss KM (1998) Linkage disequilibrium mapping of complex disease: Fantasy or reality? *Current Opinion in Biotechnology* 9:578–594
- Thomas A (2005) Characterizing allelic associations from unphased diploid data by graphical modeling. *Genetic Epidemiology* 29:23–35
- (2007) Towards linkage analysis with markers in linkage disequilibrium by graphical modelling. *Human Heredity* : in press
- Thompson EA (2000) Statistical Inferences from Genetic Data on Pedigrees, vol. 6 of *NSF-CBMS Regional Conference Series in Probability and Statistics*. Institute of Mathematical Statistics, Beachwood, OH
- (2005) MCMC in the analysis of genetic data on pedigrees. In F Liang, JS Wang, W Kendall, eds., *Markov Chain Monte Carlo: Innovations and Applications*, 183–216. World Scientific Co Pte Ltd, Singapore
- (2006) Uncertainty in inheritance: Assessing evidence for linkage. Technical report #498, Department of Statistics, University of Washington
- Thompson EA, Geyer CJ (2007) Fuzzy p-values in latent variable problems. *Biometrika* : in press

- Thompson EA, Guo SW (1991) Evaluation of likelihood ratios for complex genetic models. *IMA Journal of Mathematics Applied in Medicine and Biology* 8:149–169
- Tibshirani R (1996) Regression shrinkage and selection via the LASSO. *Journal of the Royal Statistical Society B* 58:267–288
- Tong L, Thompson EA (2007) Multilocus lod scores in large pedigrees: Combination of exact and approximate calculations. Submitted
- Tuzun E, Sharp AJ, Bailey JA, Kaul R, Morrison VA, Pertz LM, Haugen E, Hayden H, Albertson D, Pinkel D, Olson MV, Eichler EE (2005) Fine-scale structural variation of the human genome. *Nature Genetics* 37:727–732
- Ueda T, Sato T, Hidema J, Hirouchi T, Yamamoto K, Kumagai T, Yano M (2005) qUVR-10, a major quantitative trait locus for ultraviolet-B resistance in rice, encodes cyclobutane pyrimidine dimer photolyase. *Genetics* 171:1941–1950
- van Heel DA, McGovern DPB, Cardon LR, Dechairo BM, Lench NJ, Carey AH, Jewell DP (2002) Fine mapping of the IBD1 locus did not identify Crohn disease-associated NOD2 variants: Implications for complex disease genetics. *American Journal of Human Genetics* 111:253–259
- Verzilli CJ, Stallard N, Whittaker JC (2006) Bayesian graphical models for genomewide association studies. *American Journal of Human Genetics* 79:100–112
- Viallefont V, Raftery AE, Richardson S (2001) Variable selection and Bayesian model averaging in case-control studies. *Statistics in Medicine* 20:3215–3230
- Vogelstein B, Kinzler KW (2004) Cancer genes and the pathways they control. *Nature Medicine* 10:789–799
- Walsh DM, Selkoe DJ (2004) Oligomers in the brain: The emerging role of soluble protein aggregates in neurodegeneration. *Protein and Peptide Letters* 11:213–228
- Wermuth N (1976) Model search among multiplicative models. *Biometrics* 32:253–263
- Whittaker J (1984) Fitting all possible decomposable and graphical models to multiway contingency tables. In T Havranek, Z Sidak, M Novak, eds., *COMPSTAT 1984 – Sixth Symposium on Computational Statistics*, 401–406. Physica-Verlag Ges.m.b.H
- Whittemore A, Halpern J (1994) A class of tests for linkage using affected pedigree members. *Biometrics* 50:118–127
- Wijsman EM (2005) Gene mapping and the transition from STRPs to SNPs. In LB Jorde, PFR Little, MJ Dunn, S Subramaniam, eds., *Encyclopedia of Genetics, Genomics, Proteomics and Bioinformatics*. Wiley & Sons, New York
- Wijsman EM, Amos CI (1997) Genetic analysis of simulated oligogenic traits in nuclear and extended pedigrees: Summary of GAW10 contributions. *Genetic Epidemiology* 14:719–735
- Wijsman EM, Rothstein JH, Thompson EA (2006) Multipoint linkage analysis with many multiallelic or dense diallelic markers: MCMC provides practical approaches for genome scans on general pedigrees. *American Journal of Human Genetics* 79:846–858
- Wilcox MA, Pugh EW, Zhang H, Zhong X, Levinson DF, Kennedy GC, Wijsman EM (2005) Comparison of single-nucleotide polymorphisms and microsatellite markers for linkage analysis in the COGA and simulated data sets for Genetic Analysis Workshop 14: Presentation groups 1, 2, and 3. *Genetic Epidemiology* 29 (Suppl 1):S7–S28
- Williams JT, Van Eerdewegh P, Almasy L, Blangero J (1999) Joint multipoint linkage analysis of multivariate qualitative and quantitative traits. I. Likelihood formulation and simulation results. *American Journal of Human Genetics* 65:1134–1147

- Wong KK, deLeeuw RJ, Dosanjh NS, Kimm LR, Cheng Z, Horsman DE, MacAulay C, Ng RT, Brown CJ, Eichler EE, Lam WL (2007) A comprehensive analysis of common copy-number variations in the human genome. *American Journal of Human Genetics* 80:91–104
- Yang ZL, Camp NJ, Sun H, Tong ZZ, Gibbs D, Cameron DJ, Chen HY, Zhao Y, Pearson E, Li X, Chien J, DeWan A, Harmon J, Bernstein PS, Shridhar V, Zabriskie NA, Hoh J, Howes K, Zhang K (2006) A variant of the HTRA1 gene increases susceptibility to age-related macular degeneration. *Science* 314:992–993
- Yu CE, Dawson G, Munson J, D’Souza I, Osterling J, Estes A, Leutenegger AL, Flodman P, Smith M, Raskind WH, Spence MA, McMahan W, Wijsman EM, Schellenberg GD (2002) Presence of large deletions in kindreds with autism. *American Journal of Human Genetics* 71:100–115
- Yu CE, Devlin B, Galloway N, Loomis E, Schellenberg GD (2004) ADLAPH: A molecular haplotyping method based on allele-discriminating long-range PCR. *Genomics* 84:600–612
- Zeng ZB (1994) Precision mapping of quantitative trait loci. *Genetics* 136:1457–1468