

Publication list; Keith Humphreys

1. Li J, Humphreys K, Darabi H, Rosin G, Hannelius U, Heikkinen T, Aittomäki K, Blomqvist C, Pharoah PDP, Dunning AM, Ahmed S, Hooning MJ, Hollestelle A, Oldenburg RA, Alfredsson L, Palotie A, Peltonen-Palotie L, Irwanto A, Low HQ, Teoh GHK, Thalamuthu A, Kere J, D'Amato M, Easton DF, Nevanlinna H, Liu J, Czene K, Hall P. A genome-wide association scan on ER-negative breast cancer. *Breast Cancer Research*. 2010, 12:R93 (9 November 2010).
2. Darabi H, Czene K, Wedrén S, Li Y, Liu J, Hall P and Humphreys K. Genetic Variation in the Androgen Estrogen Conversion Pathway in Relation to Breast Cancer prognosticators. *Breast Cancer Research and Treatment*. 2010 Oct 19. [Epub ahead of print].
3. Fletcher O, Johnson N, Dos Santos Silva I, Orr N, Ashworth A, Nevanlinna H, Heikkinen T, Aittomäki K, Blomqvist C, Burwinkel B, Bartram CR, Meindl A, Schmutzler RK, Cox A, Brock I, Elliott G, Reed MW, Southey MC, Smith L, Spurdle AB, Hopper JL, Couch FJ, Olson JE, Wang X, Fredericksen Z, Schürmann P, Waltes R, Bremer M, Dörk T, Devilee P, van Asperen CJ, Tollenaar RA, Seynaeve C, Hall P, Czene K, Humphreys K, Liu J, Ahmed S, Dunning AM, Maranian M, Pharoah PD, Chenevix-Trench G; for the kConFab Investigators and the AOCS Group, Beesley J, Bogdanova NV, Antonenkova NN, Zalutsky IV, Anton-Culver H, Ziogas A, Brauch H, Ko YD, Hamann U; for the GENICA Consortium, Fasching PA, Strick R, Ekici AB, Beckmann MW, Giles GG, Severi G, Baglietto L, English DR, Milne RL, Benítez J, Arias JI, Pita G, Nordestgaard BG, Bojesen SE, Flyger H, Kang D, Yoo KY, Noh DY, Mannermaa A, Kataja V, Kosma VM, García-Closas M, Chanock S, Lissowska J, Brinton LA, Chang-Claude J, Wang-Gohrke S, Broeks A, Schmidt MK, van Leeuwen FE, Van't Veer LJ, Margolin S, Lindblom A, Humphreys MK, Morrison J, Platte R, Easton DF, Peto J; for the Breast Cancer Association Consortium. (2010) Missense Variants in ATM in 26,101 Breast Cancer Cases and 29,842 Controls. *Cancer Epidemiol Biomarkers Prev*; 19; 2143-51
4. Jingmei Li, Keith Humphreys, Tuomas Heikkinen, Kristiina Aittomäki, Carl Blomqvist, Paul D. P. Pharoah, Alison M. Dunning, Shahana Ahmed, Maartje J. Hooning, John W. M. Martens, Ans M. W. van den Ouweland, Lars Alfredsson, Aarno Palotie, Leena Peltonen-Palotie, Astrid Irwanto, Hui Qi Low, Garrett H. K. Teoh, Anbupalam Thalamuthu, Douglas F. Easton, Heli Nevanlinna, Jianjun Liu, Kamila Czene, Per Hall. A combined analysis of genome-wide association studies in breast cancer. *Breast Cancer Research and Treatment*. 2010 Sep 25. [Epub ahead of print]
5. Ekström Smedby K, Eloranta S, Duvefeldt K, Hjalgrim H, Humphreys K, Melbye M and Chang ET. Vitamin D receptor genotypes, ultraviolet radiation exposure and risk of non-Hodgkin lymphoma. *American Journal of Epidemiology*. Accepted.
6. Bergman S, Söderqvist S, Peyrard-Janvid M, Matsson H, Thorell L, Humphreys K, Kere J and Klingberg T et al. Gains in fluid intelligence after training non-verbal reasoning in 4-year-old children – a controlled, randomized study. *Developmental Science*. Accepted.
7. Söderqvist S, McNab F, Peyrard-Janvid M, Matsson H, Humphreys K, Kere J and Klingberg T. The SNAP25 gene is linked to working memory capacity and maturation of the posterior cingulate cortex during childhood. *Biological Psychiatry*. Accepted.

8. Leu M, Humphreys K, Surakka I, Rehnberg E, Muilu, J, Rosenström P, Almgren P, Jääskeläinen J, Lifton RP, Kyvik KO, Kaprio J, Pedersen NL, Palotie A, Hall P, Grönberg H, Groop L, Peltonen L, Palmgren J and Rippatti S. A Nordic pool and portal for genome-wide control data; Implications for association studies on a Nordic scale. *European Journal of Human Genetics*. 2010 Jul 28. [Epub ahead of print]
9. Grunewald M, Humphreys K and Hössjer O. (2010) A stochastic EM type algorithm for parameter estimation in models with continuous outcomes, under complex ascertainment. *International Journal of Biostatistics*. Vol.6: Iss. 1, Article 23.
10. Conde L, Halperin E, Akers NK, Brown KM, Smedby KE, Rothman N, Nieters A, Slager SL, Brooks-Wilson A, Agana L, Riby J, Liu J, Adami HO, Darabi H, Hjalgrim H, Low HQ, Humphreys K, Melbye M, Chang ET, Glimelius B, Cozen W, Davis S, Hartge P, Morton LM, Schenk M, Wang SS, Armstrong B, Krickler A, Milliken S, Purdue MP, Vajdic CM, Boyle P, Lan Q, Zahm SH, Zhang Y, Zheng T, Becker N, Benavente Y, Boffetta P, Brennan P, Butterbach K, Cocco P, Foretova L, Maynadié M, de Sanjosé S, Staines A, Spinelli JJ, Achenbach SJ, Call TG, Camp NJ, Glenn M, Caporaso NE, Cerhan JR, Cunningham JM, Goldin LR, Hanson CA, Kay NE, Lanasa MC, Leis JF, Marti GE, Rabe KG, Rassenti LZ, Spector LG, Strom SS, Vachon CM, Weinberg JB, Holly EA, Chanock S, Smith MT, Bracci PM, Skibola CF (2010). Genome-wide association study identifies novel HLA-DQB1 risk alleles specific to follicular lymphoma. *Nature Genetics*. 42: 661-4.
11. Sjölander A, Vansteelandt S. and Humphreys K. (2010) A principal stratification approach to assess the differences in prognosis between cancers caused by hormone replacement therapy and by other factors. *International Journal of Biostatistics*. Vol.6: Iss. 1, Article 20.
12. Low YL, Li Y, Humphreys K, Thalamuthu A, Li Y, Darabi H, Wedrén S, Bonnard C, Czene K, Iles M, Heikkinen T, Aittomäki K, Blomqvist C, Nevanlinna H, Hall P, Liu ET and Liu J. (2010) Multi-variant Pathway Association Analysis Reveals the Importance of Genetic Determinants of Estrogen Metabolism in Breast and Endometrial Cancer Susceptibility. *PLoS Genetics*. 6:e1001012.
13. Fernberg P, Chang ET, Duvefelt K, Hjalgrim H, Eloranta S, Sørensen KM, Porwit A, Humphreys K, Melbye M, Ekström Smedby K. (2010) Genetic variation in chromosomal translocation breakpoint and immune function genes and risk of non-Hodgkin lymphoma. *Cancer Causes Control*. 21: 759-769.
14. Li J, Humphreys K, Eriksson L, Czene K, Jiu J and Hall P. (2010) Effects of childhood body size on breast cancer tumour characteristics. *Breast Cancer Research*. 12: R23.
15. Li J, Eriksson L, Humphreys K, Czene K, Liu J, Tamimi R, Lindström S, Hunter DJ, Vachon C, Couch F, Christopher S, Laggiou P and Hall P. (2010) Genetic variation in the estrogen metabolic pathway and mammographic density as an intermediate phenotype of breast cancer. *Breast Cancer Research*. 12: R19..
16. Bergman Nutley S, Söderqvist S, Bryde S, Humphreys K and Klingberg T (2010) Measuring working memory capacity with greater precision in the lower capacity ranges. *Developmental Neuropsychology*. 35, 81-95.
17. Hössjer O, Hartman L and Humphreys K. (2009) Ancestral recombination graphs under nonrandom ascertainment with applications to gene mapping. *Statistical Applications of Genetics and Molecular Biology*. Vol. 8. Issue 1.

18. Einarsdottir K, Darabi H, Czene K, Li Y, Low YL, Li YQ, Bonnard C, Wedren S, Liu ET, Hall P, Liu J and Humphreys K (2009) Common Genetic variability in ESR1 and EGF in relation to Endometrial cancer risk and survival. *Br. J. Cancer.* 100:1358-64.
19. Eike, M. C., Becker, T., Humphreys, K., Olsson, M., and B. A. Lie. (2009) Conditional analyses on the T1DGC MHC dataset: novel associations with type 1 diabetes around HLA-G and confirmation of HLA-B. *Genes Immun.* 10:56-67.
20. Ripatti S, Becker T, Bickebller H, Dominicus A, Fischer C, Humphreys K, Jonasdottir G, Moreau Y, Olsson M, Ploner A, Sheehan N, Van Steen K, Baur M, van Duijn C, Palmgren J. (2009) GENESTAT: an information portal for design and analysis of genetic association studies. *Eur J Hum Genet.* 17: 533-6.
21. Eike MC, Humphreys K, Becker T, Olsson M, Lie BA (2009) Three microsatellites from the T1DGC MHC data set show highly significant association with type 1 diabetes, independent of the HLA-DRB1, -DQA1 and -DQB1 genes. *Diabetes Obes Metab.* 11 Suppl 1:17-24.
22. Hartman L, Hössjer and Humphreys K. (2009) Using Identity-By-Descent probabilities for genetic fine mapping in population based samples, via spatial smoothing of haplotype effects. *Computational Statistics and Data Analysis.* 53: 1802-17.
23. Sjölander A, Humphreys K, Vansteelandt S, Bellocco R and Palmgren J. (2009) Sensitivity analysis for principal stratum direct effects. *Biometrics.* 65:514-20.
24. Garcia-Closas M, Hall P, Nevanlinna H, Pooley K, Morrison J, Richesson DA, Bojesen SE, Nordestgaard BG, Axelsson CK, Arias JI, Milne RL, Ribas G, González-Neira A, Benítez J, Zamora P, Brauch H, Justenhoven C, Hamann U, Ko YD, Bruening T, Haas S, Dörk T, Schürmann P, Hillemanns P, Bogdanova N, Bremer M, Karstens JH, Fagerholm R, Aaltonen K, Aittomäki K, von Smitten K, Blomqvist C, Mannermaa A, Uusitupa M, Eskelinen M, Tengström M, Kosma VM, Kataja V, Chenevix-Trench G, Spurdle AB, Beesley J, Chen X, Australian Ovarian Cancer Management Group, Kathleen Cuninghame Foundation Consortium For Research Into Familial Breast Cancer, Devilee P, van Asperen CJ, Jacobi CE, Tollenaar RA, Huijts PE, Klijn JG, Chang-Claude J, Kropp S, Slanger T, Flesch-Janys D, Mutschelknauss E, Salazar R, Wang-Gohrke S, Couch F, Goode EL, Olson JE, Vachon C, Fredericksen ZS, Giles GG, Baglietto L, Severi G, Hopper JL, English DR, Southey MC, Haiman CA, Henderson BE, Kolonel LN, Le Marchand L, Stram DO, Hunter DJ, Hankinson SE, Cox DG, Tamimi R, Kraft P, Sherman ME, Chanock SJ, Lissowska J, Brinton LA, Peplonska B, Klijn JG, Hooning MJ, Meijers-Heijboer H, Collee JM, van den Ouweland A, Uitterlinden AG, Liu J, Lin LY, Yuqing L, Humphreys K, Czene K, Cox A, Balasubramanian SP, Cross SS, Reed MW, Blows F, Driver K, Dunning A, Tyrer J, Ponder BA, Sangrajrang S, Brennan P, McKay J, Odefrey F, Gabrieau V, Sigurdson A, Doody M, Struewing JP, Alexander B, Easton DF, Pharoah PD. (2008) Heterogeneity of breast cancer associations with five susceptibility loci by clinical and pathological characteristics. *PLoS Medicine* 4(4):e1000054.
25. Einarsdottir K, Darabi H, Li Y, Low YL, Li Y, Bonnard C, Sjölander A, Czene K, Wedren S, Liu ET, Hall P, Humphreys K, and Liu J (2008) ESR1 and EGF Genetic variation in relation to breast cancer risk and survival. *Breast Cancer Research* 10(1) R:15.

26. Wedren S, Lovmar L, Humphreys K, Magnusson C, Melhus H, Syvnen AC, Kindmark A, Landegren U, Fermr ML, Stiger F, Persson I, Baron JA, Weiderpass E. (2008) Estrogen receptor alpha gene polymorphism and endometrial cancer risk--a case-control study. *BMC Cancer* 8:322
27. Jonasdottir G, Becker T, Humphreys K, Palmgren J. (2008) Testing association in the presence of linkage using the GRE and multiple markers. *Genetic Epidemiology*. 32; 425-33.
28. Sjölander A, Humphreys K, Palmgren J (2008). On informative detection bias in screening studies. *Statistics in Medicine*. 27(14):2635-50
29. Wedren S, Magnusson C, Humphreys K, Malhus H, Kindmark A, Stiger F, Branting M, Persson I, Baron J, Weiderpass E. (2007) Androgen and Vitamin D Receptor microsatellites and postmenopausal breast cancer. *Cancer Epidemiol. Biomarkers Prev*. 16; 1775-17783.
30. Nordman S, Abulaiti A, Hilding A Langberg E-W, Humphreys K, Östenson CG, Efendic S and Gu HF (2008). Genetic Variation of the Adenylyl Cyclase 3 (AC3) Locus and Its Influences to Type 2 Diabetes and Obesity Susceptibility in Swedish Men. *International Journal of Obesity*. 32:407-12.
31. Sjölander A, Hössjer O, Werner-Hartman L, Humphreys K (2007). Fine mapping of disease genes using tagging SNPs. *Annals of Human Genetics* 71: 815-827.
32. Jonasdottir G, Humphreys K, Palmgren J. (2007) A powerful score for testing association in the presence of linkage using binary data. *Genetic Epidemiology*. 31: 528-540.
33. Einarsdottir K, Humphreys K, Bonnard C, Li Y, Li Y, Chia KS, Liu ET, Hall P, Liu J, Wedren S. (2007) Effect of ATM, ERBB2 and CHEK2 tagSNPs and haplotypes on endometrial cancer risk. *Human Molecular Genetics*. 16. 154-1
34. Einarsdottir K, Rosenberg, LU, Humphreys K, Bonnard C, Palmgren J, Li Y, Li Y, Chia KS, Liu ET, Hall P, Liu J, Wedren S. (2006) Comprehensive analysis of the ATM, ERBB2 and CHEK2 genes in relation to breast cancer tumour characteristics and survival. *Breast Cancer Research*. 8. R67.
35. Dahlman I, Nilsson M, Jiao H, Hoffstedt J, Lindgren CM, Humphreys K, Kere J, Gustafsson J-A, Arner P and Dahlman-Wright K. (2006) Liver X receptor gene polymorphisms and adipose tissue expression levels in obesity. *Pharmacogenetics and Genomics*. 16, 881-889.
36. Einarsdottir K, Humphreys K, Bonnard C, Palmgren J, Iles M, Sjölander A, Li Y, Chia KS, Liu E, Hall P, Liu J and Wedren S (2006) Linkage Disequilibrium Mapping of CHEK2: Common Variation and Breast Cancer Risk. *PLoS Medicine*. 3, e168.
37. Ekström-Smedby K, Lindgren C, Hjalgrim H, Humphreys K, Schllkopf C, Chang ET, Roos G, Ryder LP, Falk KI, Palmgren J, Kere J, Melbye M, Glimelius B, Adami H-O. (2006) Variation in DNA repair genes ERCC2, XRCC1 and XRCC3 and risk of follicular lymphoma. *Cancer Epidemiol. Biom. Prev*. 15: 258-265.
38. Einarsdottir K, Rylander-Rudqvist T, Humphreys K, Ahlberg S, Jonasdottir G, Weiderpass E, Chia KS, Ingelman-Sundberg M, Persson I, Liu J, Hall P, Wedren S. (2005) CYP17 gene polymorphism in relation to breast cancer risk: a case-control study. *Breast Cancer Research* 7:R890-R896.

39. Jonasdottir G, Palmgren J and Humphreys K (2005) Analysis of binary traits. Testing association in the presence of linkage. *BMC Genetics* 6 Suppl 1:S92
40. Humphreys, K and Iles, M. (2005) Fine scale mapping in case-control samples using locus scoring and haplotype sharing methods. *BMC Genetics* 6 Suppl 1:S74.
41. Wedren S, Lovmar L, Humphreys K, Magnusson C, Melhus H, Syvanen AC, Kindmark A, Landegren U, Fermer ML, Stiger F, Persson I, Baron J, Weiderpass E. (2004) Oestrogen receptor alpha gene haplotype and postmenopausal breast cancer risk: a case control study. *Breast Cancer Research* 6: 437-449.
42. Pawitan Y, Bjöhlke J, Wedren S, Humphreys K, Skoog L, Huang F, Amler L, Shaw P, Hall P, Bergh J (2004) Gene expression profiling using Cox regression. *Statistics in Medicine* 23: 1767-1780.
43. Humphreys, K and Palmgren, J (2004) Ascertainment corrected score tests for haplotype trait association (Abstract) *Genetic Epidemiology* 27: 276.
44. Gu HF, Abulaiti A, Östenson C-G, Humphreys K, Wahlestedt C, Brookes AJ, Efendic S. (2004) Single nucleotide polymorphisms in the proximal promoter region of the Adiponectin (APM1) gene are associated with type 2 diabetes in Swedish Caucasians. *Diabetes* 53: 31-35.
45. Rylander-Rudqvist T, Wedren S, Granath F, Humphreys K, Ahlberg S, Weiderpass E, Oscarson M, Ingelman-Sundberg, Persson I (2003) Cytochrome P450 1B1 gene polymorphisms and postmenopausal breast cancer risk. *Carcinogenesis* 24: 1533-1539.
46. Humphreys, K. and Titterton, D.M. (2003) Variational approximations for categorical causal modelling with latent variables. *Psychometrika*, 68: 391-412.
47. Hall, P., Humphreys, K. and Titterton, D.M. (2002). On the adequacy of variational lower bound functions for likelihood-based inference in Markovian models with missing values. *Journal of the Royal Statistical Society, B*, 64, 549-564.
48. Humphreys, K. and Titterton, D.M. (2001). Some examples of recursive variational approximations for Bayesian inference. In M. Opper and D. Saad (Eds.) *Advanced Mean Field Methods - Theory and Practice* (pp. 179-195). MIT Press.
49. Humphreys, K. and Titterton, D.M. (2000). Approximate Bayesian inference for simple mixtures. In J.G. Bethlehem and P.G.M. van der Heijden (Eds.) *COMPSTAT: Proceeding in computational statistics 2000* (pp. 331-336). Heidelberg: Physica Verlag.
50. Humphreys, K. and Titterton, D.M. (2000). Improving the mean field approximation in belief networks using Bahadur's reparameterisation of the multivariate binary distribution. *Neural Processing Letters*, 12, 183-197.
51. Humphreys, K. and Janson, H. (2000). Latent transition analysis with covariates, nonresponse, summary statistics and diagnostics. *Multivariate Behavioral Research*, 35, 89-118.
52. Humphreys, K. (2000). Latent variable models and factor analysis. Book review. (Authors Bartholomew, D.J. and Knott, M.). *Statistical Methods in Medical Research*, 9, 415-6.

53. Humphreys, K. and Titterton, D.M. (1999). The exploration of new methods for learning in binary Boltzmann machines. In D. Heckerman and J. Whittaker (Eds.) *Proceedings of the Seventh International Workshop on Artificial Intelligence and Statistics* (Pp. 209-214). Morgan Kaufmann, San Francisco.
54. Skinner, C.J. and Humphreys, K. (1999). Weibull regression for lifetimes measured with error. *Lifetime data analysis*, 5, 23-37.
55. Humphreys, K. (1998). The latent Markov chain with multivariate random effects: an evaluation of instruments measuring labour market status in the British Household Panel Study. *Sociological Methods and Research*, 26, 3, 269-299.
56. Pfeiffermann, D., Skinner, C.J. and Humphreys, K. (1998). The estimation of gross flows in the presence of measurement error using auxiliary variables. *Journal of the Royal Statistical Society, A*, 161, 1, 13-32.
57. Humphreys, K. (1997). Classification error adjustments for female labour force transitions using a latent Markov chain with multivariate random effects. In J. Rost and R. Langeheine (Eds.) *Applications of latent trait and latent class models in the Social Sciences* (Pp. 370-379). Waxmann Munster, New York.
58. Humphreys, K. and Skinner, C.J. (1997). Instrumental Variable Estimation of Gross Flows in the presence of measurement error. *Survey Methodology*, 23, 1, 53-60.
59. Humphreys, K. and Skinner, C.J. (1994). A review of PANMARK. *Journal of the Royal Statistical Society, C*, 43, 415-7.
60. Humphreys, K. and Carr-Hill, R.A. (1991). Area variations in health outcomes: Artefact or ecology? *International Journal of Epidemiology*, 20, 1, 251-8.
61. Skinner, C.J., Humphreys, K. and Holt, D. (1991). Analysis of survival times recorded with error. *Bulletin of the International Statistical Institute*. Contributed Papers. 54(2), 632-3.
62. Humphreys, K. and Carr-Hill, R.A. (1990). *Area variations in Health outcomes. In Lifestyle, health and health promotion: proceedings of a symposium on health and lifestyle*. HPRT, Cambridge.
63. Carr-Hill, R.A., Humphreys, K. and McIver, S. (1987). A customised view of public satisfaction. *The Health Services Journal*, 28 May 1987, pp. 614-5.