Publications
Matti Pirinen
23 - Dec - 2017

- * denotes equal contribution, i.e., a shared first/second authorship
- H-index = 23 (Web of Science, 23-Dec-2017), 29 (Google Scholar, 23-Dec-2017)

A. Peer-reviewed scientific publications
A1. Original research publications

1. Almangush A, **Pirinen M**, Heikkinen I, Mäkitie AA, Salo T, Leivo I **(2017)**:
   Tumour budding in oral squamous cell carcinoma: a meta-analysis.
   British Journal of Cancer doi:10.1038/bjc.2017.425

2. Benner C, Havulinna AS, Jarvelin MR, Salomaa V, Ripatti S, **Pirinen M (2017)**:
   Prospects of Fine-Mapping Trait-Associated Genomic Regions by Using Summary Statistics from Genome-wide Association Studies.
   American Journal of Human Genetics 101: 539-551.

3. Kerminen S, Havulinna AS, Hellenthal G, Martin AR, Sarin AP, Perola M, Palotie A, Salomaa V, Daly MJ, Ripatti S, **Pirinen M (2017)**:
   Fine-Scale Genetic Structure in Finland.
   G3-Genes Genomes Genetics 7: 3459-3468.

4. **Pirinen M**, Benner C, Marttinen P, Jarvelin MR, Rivas MA, Ripatti S **(2017)**:
   biMM: efficient estimation of genetic variances and covariances for cohorts with high-dimensional phenotype measurements.
   Bioinformatics 33: 2405-2407.

5. Chheda H, Palta P, **Pirinen M**, McCarthy S, Walter K, Koskinen S, Salomaa V, Daly M, Durbin R, Palotie A, Aittokallio T, Ripatti S **(2017)**:
   Whole-genome view of the consequences of a population bottleneck using 2926 genome sequences from Finland and United Kingdom.
   European Journal of Human Genetics 25: 477-484.

6. Wang L, Ko ER, Gilchrist JJ, Pittman KJ, Rautanen A, **Pirinen M**, Thompson JW, Dubois LG, Langley RJ, Jaslow SL, Salinas RE, Rouse DC, Moseley MA, Mwarumba S, Njuguna P, Mturi N, Williams TN, Scott JAG, Hill AVS, Woods CW, Ginsburg GS, Tsalik EL, Ko DC **(2017)**:
   Human genetic and metabolite variation reveals that methylthioadenosine is a prognostic biomarker and an inflammatory regulator in sepsis.
   Science Advances 3: e1602096.

7. Tikkanen E, **Pirinen M**, Sarin AP, Havulinna AS, Männistö S, Saltevo J, Lokki ML, Sinisalo J, Lundqvist A, Jula A, Salomaa V, Ripatti S **(2016)**:
Genetic support for the causal role of insulin in coronary heart disease.
Diabetologia 59: 2369-2377.

8. Gillberg J, Marttinen P, **Pirinen M**, Kangas AJ, Soininen P, Ali M, Havulinna AS, Järvelin MR, Ala-Korpela M, Kaski S **(2016)**:
Multiple output regression with latent noise.
Journal of Machine Learning Research 17: 4170-4204.

9. Sieberts SK*, Zhu F*, García-García J*, Stahl E, Pratap A, Pandey G, Pappas D, Aguilar D, Anton B, Bonet J, Eksi R, Fornés O, Guney E, Li H, Marín MA, Panwar B, Planas-Iglesias J, Poglayen D, Cui J, Falcao AO, Suver C, Hoff B, Balagurusamy VSK, Dillenberger D, Neto EC, Norman T, Aittokallio T, Ammad-ud-din M, Azencott CA, Bellón V, Boeva V, Bunte K, Chheda H, Cheng L, Corander J, Dumontier M, Goldenberg A, Gopalacharyulu P, Hajiloo M, Hidru D, Jaiswal A, Kaski S, Khalfaoui B, Khan SA, Kramer ER, Marttinen P, Mezlini AM, Molparia B, **Pirinen M** et al. **(2016)**:
Crowdsourced assessment of common genetic contribution to predicting anti-TNF treatment response in rheumatoid arthritis.
Nature Communications 7: 12460.

10. Rautanen A*, **Pirinen M***, Mills TC, Rockett KA, Strange A, Ndungu AW, Naranbhai V, Gilchrist JJ, Bellenguez C, Freeman C, Band G, Bumpstead SJ, Edkins S, Giannoulatou E, Gray E, Dronov S, Hunt SE, Langford C, Pearson RD, Su Z, Vukcevic D, Macharia AW, Uyoga S, Ndila C, Mturi N, Njuguna P, Mohammed S, Berkley JA, Mwangi I, Mwarumba S, Kitsao BS, Lowe BS, Morpeth SC, Khandwalla I; Kilifi Bacteraemia Surveillance Group, Blackwell JM, Bramon E, Brown MA, Casas JP, Corvin A, Duncanson A, Jankowski J, Markus HS, Mathew CG, Palmer CN, Plomin R, Sawcer SJ, Trembath RC, Viswanathan AC, Wood NW, Deloukas P, Peltonen L, Williams TN, Scott JA, Chapman SJ, Donnelly P, Hill AV, Spencer CC **(2016)**:
Polymorphism in a lincRNA associates with a doubled risk of Pneumococcal Bacteremia in Kenyan children.
American Journal of Human Genetics 98: 1092-1100.

11. Ripatti P, Rämö JT, Söderlund S, Surakka I, Matikainen N, **Pirinen M**, Pajukanta P, Sarin AP, Service SK, Laurila PP, Ehnholm C, Salomaa V, Wilson RK, Palotie A, Freimer NB, Taskinen MR, Ripatti S **(2016):**
The Contribution of GWAS loci in Familial Dyslipidemias.
PLoS Genetics 12: e1006078.

12. Kettunen J, Demirkan A, Würtz P, Draisma HHM, Haller T, Rawal R, Vaarhorst A, Kangas AJ, Lyytikäinen LP, **Pirinen M**, Pool R, Sarin AP, Soininen P, Tukiainen T, Wang Q, Tiainen M, Tynkkynen T, Amin N, Zeller T, Beekman M, Deelen J, van Dijk KW, Esko T, Hottenga JJ, van Leeuwen EM, Lehtimäki T, Mihailov E, Rose RJ, de Craen AJM, Gieger C, Kähönen M, Perola M, Blankenberg S, Savolainen MJ, Verhoeven A, Viikari J, Willemsen G, Boomsma DI, van Duijn CM, Eriksson J, Jula A, Järvelin MR, Kaprio J, Metspalu A, Raitakari O, Salomaa V, Slagboom PE,

Waldenberger M, Ripatti S, Ala-Korpela M **(2016)**:
Genome-wide study for circulating metabolites identifies 62 loci and reveals novel systemic effects of LPA.
Nature Communications 7:11122.

13. Cichonska A, Rousu J, Marttinen P, Kangas AJ, Soininen P, Lehtimäki T, Raitakari OT, Järvelin MR, Salomaa V, Ala-Korpela M, Ripatti S, **Pirinen M (2016)**:
metaCCA: summary statistics-based multivariate meta-analysis of genome-wide association studies using canonical correlation analysis.
Bioinformatics 32: 1981-1989.

14. Benner C, Spencer CCA, Havulinna A, Salomaa V, Ripatti S, **Pirinen M (2016)**:
FINEMAP: Efficient variable selection using summary data from genome-wide association studies.
Bioinformatics 32: 1493-1501.

15. Rivas MA, **Pirinen M***, Conrad DF*, Lek M*, Tsang EK, Karczewski KJ, Maller JB, Kukurba KR, DeLuca D, Fromer M, Ferreira PG, Smith KS, Zhang R, Zhao F, Banks E, Poplin R, Ruderfer D, Purcell SM, Tukiainen T, Minikel EV, Stenson PD, Cooper DN, Huang KH, Sullivan TJ, Nedzel T, the GTEx Consortium, the Geuvadis Consortium, Bustamante CD, Li JB, Daly MJ, Guigo R, Donnelly P, Ardlie K, Sammeth M, Dermitzakis E, McCarthy MI, Montgomery SB, Lappalainen T, MacArthur DG **(2015):**
Impact of predicted protein-truncating genetic variants on the human transcriptome.
Science 348: 666-669.

16. **Pirinen M**, Lappalainen T, Zaitlen NA, GTEx Consortium, Dermitzakis ET, Donnelly P, McCarthy MI, Rivas MA **(2015):**
Assessing allele specific expression across multiple tissues from RNA-seq read data.
Bioinformatics 31:2497-2504.

17. Baran Y, Subramaniam M, Biton A, Tukiainen T, Tsang EK, Rivas MA ,**Pirinen M**, Gutierrez-Arcelus M, Smith KS, Kukurba KR, Zhang R, Eng C, Torgerson DG, Urbanek C, GTEx Consortium, Li JB, Rodriguez-Santana JR, Burchard EG, Seibold MA, MacArthur DG, Montgomery SB, Zaitlen NA, Lappalainen T **(2015):**
The landscape of genomic imprinting across diverse adult human tissues.
Genome Research 25: 927-936.

18. Salo PP, Vaara S, Kettunen J, **Pirinen M**, Sarin AP, Huikuri H, Karhunen P, Eskola M, Nikus K, Lokki ML, Ripatti S, Havulinna A, Salomaa V, Palotie A, Nieminen MS, Sinisalo J, Perola M **(2015)**:
Genetic variants on chromosome 1p13.3 are associated with non-ST elevation myocardial infarction and the expression of DRAM2 in the Finnish populations.
PLoS ONE 10: e0140576.

19. Surakka I, Horikoshi M, Mägi R, Sarin AP, Mahajan A, Lagou V, Marullo L, Ferreira T, Miraglio B, Timonen S, Kettunen J, **Pirinen M**, Karjalainen J, et al. **(2015):**
The impact of low-frequency and rare variants on lipid levels.
Nature Genetics 47: 589-597.

20. Leslie S*, Winney S*, Hellenthal G*, Davison D, Boumertits A, Day T, Hutnik K, Royrvik EC, Cunliffe B, Lawson DJ, Falush D, Freeman C, **Pirinen M**, Myers S, Robinson M, Donnelly P, Bodmer W ***(*2015):**
Fine-scale genetic structure of the British population.
Nature 519: 309-314.

21. Rockett KA, Clark GM - 69 authors – **Pirinen M** et al. **(2014):**
Reappraisal of known malaria resistance loci in a large multi-centre study.
Nature Genetics 46: 1197-1204.

22. Davis O*, Band G*, **Pirinen M*,** Haworth CM, Meaburn EL, Kovas Y, et al. **(2014)**:
The correlation between reading and mathematics ability at age twelve has a substantial genetic component.
Nature Communications 5:4204.

23. Marttinen P, **Pirinen M**, Sarin AP, Gillberg J, Kettunen J, Surakka I, Kangas AJ, Soininen P, O'Reilly P, Kaakinen M, Kähönen M, Lehtimäki T, Ala-Korpela M, Raitakari OT, Salomaa V, Järvelin MR, Ripatti S, Kaski S. **(2014)**:
Assessing multivariate gene-metabolome associations with rare variants using Bayesian reduced-rank regression.
Bioinformatics 15:2026-2034.

24. Tukiainen T, **Pirinen M**, Sarin AP, Ladenvall C, Kettunen J, Lehtimäki T, Lokki ML, Perola M, Sinisalo J, Vlachopoulou E, Eriksson JG, Groop L, Jula A, Järvelin MR, Raitakari OT, Salomaa V, Ripatti S **(2014)**:
Chromosome X-wide association study identifies loci for fasting insulin and height and evidence for incomplete dosage compensation.
PLoS Genetics 10:e1004127.

25. Bramon E*, **Pirinen M*** et al. **(2014)**:
A genome-wide association analysis of a broad psychosis phenotype identifies three loci for further investigation.
Biological Psychiatry 75:386-397.

26. Morris DW, Pearson RD, Cormican P, Kenny EM, O'Dushlaine CT, Perreault LP, Giannoulatou E, Tropea D, Maher BS, Wormley B, Kelleher E, Fahey C, Molinos I, Bellini S, **Pirinen M**, Strange A, et al. **(2014)**:
An inherited duplication at the gene p21 Protein-Activated Kinase 7 (PAK7) is a risk factor for psychosis.

Hum Mol Genet 23:3316-3326.

27. Steinberg S, de Jong S, Mattheisen M, Costas J, Demontis D, Jamain S, Pietiläinen OP, Lin K, Papiol S, Huttenlocher J, Sigurdsson E, Vassos E, Giegling I, Breuer R, Fraser G, Walker N, Melle I, Djurovic S, Agartz I, Tuulio-Henriksson A, Suvisaari J, Lönnqvist J, Paunio T, Olsen L, Hansen T, Ingason A, **Pirinen M**, Strengman E, et al. **(2014)**:
Common variant at 16p11.2 conferring risk of psychosis.
Molecular Psychiatry 19:108-114.

28. Beecham AH, Patsopoulos NA – 142 authors – **Pirinen M** et al. **(2013)**:
Analysis of immune-related loci identifies 48 new susceptibility variants for multiple sclerosis.
Nature Genetics 45:1353-1360.

29. Lappalainen T, Sammeth M, Friedländer MR, 't Hoen PA, Monlong J, Rivas MA, Gonzàlez-Porta M, Kurbatova N, Griebel T, Ferreira PG, Barann M, Wieland T, Greger L, van Iterson M, Almlöf J, Ribeca P, Pulyakhina I, Esser D, Giger T, Tikhonov A, Sultan M, Bertier G, MacArthur DG, Lek M, Lizano E, Buermans HP, Padioleau I, Schwarzmayr T, Karlberg O, Ongen H, Kilpinen H, Beltran S, Gut M, Kahlem K, Amstislavskiy V, Stegle O, **Pirinen M**, Montgomery SB, et al. **(2013):**
Transcriptome and genome sequencing uncovers functional variation in humans.
Nature 501: 506-511.

30. Ripke S – 149 authors – **Pirinen M** et al. **(2013)**:
Genome-wide association analysis identifies 13 new risk loci for schizophrenia.
Nature Genetics 45:1150-1159.

31. Strange A, Bellenguez C, Sim X, Luben R, Hysi PG, Ramdas WD, van Koolwijk LM, Freeman C, **Pirinen M**, Su Z, et al. **(2013)**:
Genome-wide association study of intraocular pressure identifies the GLCCI1/ICA1 region as a glaucoma susceptibility locus.
Human Molecular Genetics 22:4653-4660.

32. Rivas MA*, **Pirinen M*,** Neville MJ, Gaulton KJ, Moutsianas L, GoT2D Consortium, Lindgren CM, Karpe F, McCarthy MI and Donnelly P. **(2013):**
Assessing association between protein truncating variants and quantitative traits.
Bioinformatics 29:2419-2426.

33. Band G, Le QS, Jostins L, **Pirinen M**, Kivinen K, Jallow M, Sisay-Joof F, Bojang K, Pinder M, Sirugo G, et al. **(2013)**:
Imputation-based meta-analysis of severe malaria in three African populations.
PLoS Genet 9:e1003509.

34. Fakiola M, Strange A, Cordell HJ, Miller EN, **Pirinen M**, Su Z, Mishra A, Mehrotra S, Monteiro GR, Band G, et al. **(2013)**:

Common variants in the HLA-DRB1-HLA-DQA1 HLA class II region are associated with susceptibility to visceral leishmaniasis.
Nature Genetics 45: 208-213.

35. **Pirinen M**, Donnelly P, and Spencer C **(2013)**:
Efficient computation with a linear mixed model on large-scale data sets with applications to genetic studies.
Annals of Applied Statistics 7:369-390.

36. **Pirinen M**, Donnelly P, and Spencer C **(2012)**:
Including known covariates can reduce power to detect genetic effects in case-control studies.
Nature Genetics 44:848-851.

37. Maller JB, McVean G, Byrnes J, Vukcevic D, Palin K, Su Z, Howson JM, Auton A, Myers S, Morris A, **Pirinen M**, Brown MA, et al. **(2012)**:
Bayesian refinement of association signals for 14 loci in 3 common diseases.
Nature Genetics 44:1294-1301.

38. Su Z, Gay LJ, Strange A, Palles C, Band G, Whiteman DC, Lescai F, Langford C, Nanji M, Edkins S, van der Winkel A, Levine D, Sasieni P, Bellenguez C, Howarth K, Freeman C, Trudgill N, Tucker AT, **Pirinen M**, Peppelenbosch MP, et al. **(2012)**:
Common variants at the MHC locus and at chromosome 16q24.1 predispose to Barrett's esophagus.
Nature Genetics 44:1131-1136.

39. Strange A, Riley BP, Spencer CC, Morris DW, **Pirinen M**, O'Dushlaine CT, Su Z, Maher BS, Freeman C, Cormican P, et al. **(2012)**:
*Genome-wide association study implicates HLA-C*01:02 as a risk factor at the major histocompatibility complex locus in schizophrenia.*
Biological Psychiatry 72:620-628.

40. Bellenguez C, Bevan S, Gschwendtner A, Spencer CC, Burgess AI, **Pirinen M**, Jackson CA, Traylor M, Strange A, Su Z, et al. **(2012)**:
Genome-wide association study identifies a variant in HDAC9 associated with large vessel ischemic stroke.
Nature Genetics 44:328-333.

41. Sawcer S*, Hellenthal G*, **Pirinen M***, Spencer CC*, Patsopoulos NA, Moutsianas L, Dilthey A, Su Z, et al. **(2011)**:
Genetic risk and a primary role for cell-mediated immune mechanisms in multiple sclerosis.
Nature 476:214-219.

42. Evans DM, Spencer CC, Pointon JJ, Su Z, Harvey D, Kochan G, Oppermann U, Dilthey A, **Pirinen M,** Stone MA, Appleton L, Moutsianas L, et al. **(2011)**:

Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility.
Nature Genetics 43:761-767.

43. Plagnol V, - 80 authors – **Pirinen M**, et al. **(2011)**:
A Two-Stage Meta-Analysis Identifies Several New Loci for Parkinson's Disease.
PloS Genetics 7: e1002142.

44. Spencer CC, Plagnol V, Strange A, Gardner M, Paisan-Ruiz C, Band G, Barker RA, Bellenguez C, Bhatia K, Blackburn H, Blackwell JM, Bramon E, Brown MA, Brown MA, Burn D, Casas JP, Chinnery PF, Clarke CE, Corvin A, Craddock N, Deloukas P, Edkins S, Evans J, Freeman C, Gray E, Hardy J, Hudson G, Hunt S, Jankowski J, Langford C, Lees AJ, Markus HS, Mathew CG, McCarthy MI, Morrison KE, Palmer CN, Pearson JP, Peltonen L, **Pirinen M,** Plomin R, et al. **(2011)**:
Dissection of the genetics of Parkinson's disease identifies an additional association 5' of SNCA and multiple associated haplotypes at 17q21.
Human Molecular Genetics 20:345-353.

45. Gasbarra G*, Kulathinal S*, **Pirinen M*** and Sillanpää MJ **(2011)**:
Estimating haplotype frequencies by combining data from large DNA pools with database information.
IEEE/ACM Trans Comput Biol Bioinf 8:36-44.

46. Strange A, Capon F, Spencer CC, Knight J, Weale ME, Allen MH, Barton A, Band G, Bellenguez C, Bergboer JG, Blackwell JM, Bramon E, Bumpstead SJ, Casas JP, Cork MJ, Corvin A, Deloukas P, Dilthey A, Duncanson A, Edkins S, Estivill X, Fitzgerald O, Freeman C, Giardina E, Gray E, Hofer A, Hüffmeier U, Hunt SE, Irvine AD, Jankowski J, Kirby B, Langford C, Lascorz J, Leman J, Leslie S, Mallbris L, Markus HS, Mathew CG, McLean WH, McManus R, Mössner R, Moutsianas L, Naluai AT, Nestle FO, Novelli G, Onoufriadis A, Palmer CN, Perricone C, **Pirinen M**, Plomin R et al. **(2010)**:
A genome-wide association study identifies new psoriasis susceptibility loci and an interaction between HLA-C and ERAP1.
Nature Genetics 42:985-990.

47. **Pirinen M** **(2009)**:
Estimating population haplotype frequencies from pooled SNP data using incomplete prior information.
Bioinformatics 25:3296-3302.

48. Gasbarra D, **Pirinen M**, Sillanpää MJ, and Arjas E **(2009)**:
Bayesian quantitative trait locus mapping based on reconstruction of genealogical histories.
Genetics 183:709-721.

49. **Pirinen M**, Kulathinal S, Gasbarra D and Sillanpää MJ **(2008)**:
Estimating population haplotype frequencies from pooled DNA samples using PHASE algorithm.

Genetics Research 90:509-524.

50. Gasbarra D, **Pirinen M**, Sillanpää MJ, and Arjas E **(2007)**:
Estimating Genealogies from Linked Marker Data: A Bayesian Approach.
BMC Bioinformatics 8:411.

51. Gasbarra D, **Pirinen M**, Sillanpää MJ, Salmela E and Arjas E **(2007)**:
Estimating Genealogies from Unlinked Marker Data: A Bayesian Approach.
Theor Pop Biol 72:305-322.

52. **Pirinen M** and Gasbarra D **(2006)**:
Finding Consistent Gene Transmission Patterns on Large and Complex Pedigrees.
IEEE/ACM Trans Comput Biol Bioinf 3:252-262.

B. Non-peer-reviewed scientific writings

G. Theses

G2. Master's Thesis

Analyyttisten ja koanalyyttisten ekvivalenssirelaatioiden ekvivalenssiluokkien lukumäärästä. (On the Number of Equivalence Classes of Analytic and Coanalytic Equivalence Relations in Polish Spaces.)
Master's Thesis in Mathematical logic, University of Helsinki, 2004.

G5. PhD Thesis

Bayesian Inference for Retrospective Population Genetics Models using Markov Chain Monte Carlo Methods.
Ph.D. Thesis in Statistics, University of Helsinki, 2009.
http://urn.fi/URN:ISBN:978-952-10-5602-4