

BIOGRAPHICAL SKETCH

NAME: Zhao, Jing Hua / 赵京华		POSITION TITLE: Genetic Analyst / Senior Research Associate	
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
Shandong (Medical) University	Bachelor (in Medicine)	1980-1985	Public Health
Fudan (Shanghai Medical) University	Master (in Medicine)	1985-1988	Medical Statistics
King's College London	PhD (in Statistics)	1996-2002 (part-time)	Statistical Genetics

My master courses include Linear Algebra/Design of Experiment/Medical Statistics/Multivariate Analysis/Epidemiology at Fudan, and Mathematical Statistics/Optimization Methods/Numerical Methods at Shanghai Jiaotong Universities.

A. Positions and employment

1988.9-1994.8	Research associate, Division of Sampling Survey, Center for Health Statistics Information, Ministry of Health
1994.8-1996.5	Visiting scientist, Department of Environmental Science, School of Public Health & Channing Laboratory, Medical School, Harvard University
1996.5-2002.8	PostDoc & Lecturer (2001.3-2002.8), Section of Genetic Epidemiology and Biostatistics, Division of Psychological Medicine, Institute of Psychiatry, King's College London
2002.9-2005.9	Statistician, Social and Genetic Epidemiology, Department of Epidemiology and Public Health, University College London
2005.9-2018.7	Investigator scientist in Genetics, MRC Epidemiology Unit
2018.8-	Genetic Analyst / Senior Research Associate, Cardiovascular Epidemiology Unit, Department of Public Health and Primary Care, University of Cambridge

B. Research interests

My work is human health-related research which over years includes familial aggregation, segregation, linkage, candidate genes and genomewide association studies (GWASs). The most recent is proteogenomics within the SCALLOP consortium using the Olink and mass spectrometry (MS) panels measured for the INTERVAL samples. I have also led collaborative analysis to the SCALLOP-Seq(ence, both WES and NGS) consortium and Host Genetics Initiative.

I have promoted reproducible research through CRAN (<https://cran.r-project.org>), GitHub (<https://github.com>) and websites. I developed genetic analysis package (gap), protein quantitative trait tools (pQTLtools) and curated <https://jinghuazhao.github.io/Computational-Statistics/>, <https://jinghuazhao.github.io/software-notes/> and <https://jinghuazhao.github.io/Omics-analysis/>. By closely following up developments in computational statistics, machine learning and artificial intelligence, I have made computing and omics analysis tools available from the University HPC, <https://cambridge-ceu.github.io/csd3/>, whose components include AI with BitNet, GeminiCLI, Ollama, llama.cpp, llm, featuring AI for MS data with InstaNovo & DIA-NN, molecule optimization with DrugAssist, single-cell omics with Seurat, scp, scanpy, scvi-tools, scGPT, C2S-Scale, mtDNA analysis with MToolBox, fNUMT, haplogrep as well as long-read sequencing analysis with SVanalyzer, hap.py, sniffles, truvari. Explorations in population genetics include selscan, angsd, relate, clues2 and fastsimcoal2. These are coupled with extensive experiments on various models, architectures, platforms as with libraries such as PyTorch, scikit-llm, LangChain.

C. Key publications

Zhao JH, et al. Mapping pQTLs of circulating inflammatory proteins identifies drivers of immune-mediated disease risk and novel therapeutic targets. *Nat Immunol* 2023, **24**(9):1540-1551, 10.1038/s41590-023-01588-w, <https://www.nature.com/articles/s41590-023-01588-w>.

COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* **600**:472–477 (2021); Pathak, G.A. et al. A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**:E1-E10 (2022); The Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. *Nature* **621**:E7–E26 (2023).

Zhao JH, Luan JA, Congdon P. Bayesian linear mixed model of polygenic effects. *J Stat Soft.* 2018, **85**(6):1-27. doi: 10.18637/jss.v085.i06

Zhao JH, Luan JA. Mixed modeling with whole genome data. *J Prob Stat.* 2012. doi: 10.1155/2012.485174.

Xue F, Li S, Luan J, Yuan Z, Luben RN, Khaw K-T, Wareham NJ, Loos RJF, **Zhao JH**. A latent variable partial least squares path modeling approach to regional association and polygenic effect with applications to a human obesity study. *PLoS ONE* 2012, **7**(2): e31927

Loos RJ, et al. Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nat Genet* 2008; **40**(6):768-75

Zhao JH. gap: genetic analysis package. *J Stat Soft* 2007, **23** (8):1-18. doi: 10.18637/jss.v023.i08.

Zhao JH, Brunner EJ, Kumari M, Singh-Manoux A, Hawe E, Talmud PJ, Marmot MG, Humphries SE. APOE polymorphism, socioeconomic status and cognitive function in later mid-life: The Whitehall II longitudinal study. *Soc Psychiatr and Psychiatr Epidemiol* 2005, **40**:557-563

Zhao JH, D Curtis, PC Sham. Model-free and permutation tests for allelic associations. *Hum Hered* 2000, **50**(2), 133-139.

D. Peer-reviewed publications (in chronological order)

1. **Zhao JH**. Computer software for secondary analysis of statistical data. *Chin J Health Stat* 1990, **7**:9-10.
2. **Zhao JH**. Some perspectives of SAS/STAT 6.03 on personal computers. *Chin J Health Stat* 1990, **7**:49-51.
3. Li NH, **Zhao JH**. Principal component analysis of factors affecting diabetes of the old people. *Chin J Gerontol* 1991, **11**(6): 333-334.
4. **Zhao JH**. Computer data processing for survey of total health expenditure. in Du, LX et al. eds. *The Survey of Total Health Expenditure*. Also in *The Survey of Total Health Expenditure* edited by Center for Health Statistics Information, 1993.
5. **Zhao JH**. A quick method to produce frequency table using Foxbase+. *J China Computer Users Group*. 1993 Supplement.
6. **Zhao JH**. A BASIC program for debugging Fortran code. *China Computers* 1991 Jan, and also in *Digest of Personal Computer Applications*, Kehai Hi-tech Co.

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7. **Zhao JH**. A simple method to identify your type of personal computer. *China Computers* 1991 Jun.
8. **Zhao JH**, Tan Q. Trend analysis of fertility data in Shandong province. *Chin J Health Stat* 1994, **11** (supplement).
9. **Zhao JH**, Wang CY. Internet and health statistics. *Med Info Proc Res* (Chinese). 1996. **4**(1):35-38
10. Xu X, **JH Zhao**. Ecogenetics I. *J Environ Health* 1996, **1**:43-46.
11. **Zhao JH**, Niu T. Ecogenetics II. *J Environ Health* 1996, **3**:139-144.
12. **Zhao JH**, Niu T. Ecogenetics III. *J Environ Health* 1996, **4**:189-190.
13. Sham PC, **JH Zhao**, D. Curtis. Optimal weighting scheme for affected sib-pair analysis of sibship data. *Ann Hum Genet* 1997, **61**:61-69.
14. Li T, K Xu, H Deng, G Cai, J Liu, X Liu, RA Wang, XY Xiang, **JH Zhao**, RM Murray, PC Sham, DA Collier. Association analysis of the dopamine D4 gene exon III VNTR and heroin abuse in Chinese subjects. *Mol Psychiatr* 1997, **2**:413-416.
15. Li T, HP Vallada, X Liu, T Xie, XD Tang, **JH Zhao**, MC O'Donovan, RM Murray, PC Sham, DA Collier. Analysis of CAG/CTG repeat size in Chinese subjects with schizophrenia and bipolar affective disorder using the repeat expansion detection method. *Biol Psychiatr* 1998, **44**(11):1160-5.
16. Arranz MJ, J Munro, MJ Owen, G Spurlock, PC Sham, **J Zhao**, G Kirov, DA Collier, RW Kerwin. Evidence for association between polymorphisms in the promoter and coding regions of the 5-HT_{2A} receptor gene and response to clozapine. *Mol Psychiatr* 1998, **3**:61-66.
17. Niu T, X Xu, J Rogus, Y Zhou, C Chen, J Yang, Z Fang, C Schmitz, **J Zhao**, VS Rao, K Lindpainter. Angiotensinogen gene and hypertension in Chinese. *J Clin Invest* 1998, **101**(1): 188-194.
18. **Zhao JH**, PC Sham. A method for calculating probability convolution using ternary numbers with application in the determination of twin zygosity. *Comp Stat Data Anal* 1998, **28**(2): 225-232.
19. Vallada H, D Curtis, P Sham, H Kunugi, **J Zhao**, R Murray, P McGuffin et al. A transmission disequilibrium and linkage analysis of D22S278 marker alleles in 574 families: further support for a susceptibility locus for schizophrenia at 22q12. *Schizophr Res* 1998, **32**:115-121.
20. Wright P, E Dawson, PT Donaldson, JA Underhill, PC Sham, **JH Zhao**, M Gill, S Nanko, MJ Owen, P McGuffin, RM Murray. A transmission/disequilibrium study of the DRB1*04 gene locus on chromosome 6p21.3 with schizophrenia. *Schizophr Res* 1998, **32**:75-80.
21. Ohadi M, MRA Laloz, P Sham, **J Zhao**, AM Dearlove, C Shiach, S Kinsey, M Rhodes, DM Layton. Localization of a Gene for Familial Hemophagocytic Lymphohistiocytosis at Chromosome 9q21.3-22 by Homozygosity Mapping. *Am J Hum Genet* 1999, **64**(1):165-171.
22. Abusaad I, D Mackay, **J Zhao**, P Stanford, DA Collier, IP Everall. Stereological estimation of the total number of neurons in the murine hippocampus using the optical disector. *Am J Med Genet (Neuropsychiatric Genet)* 1998, **81**(6):483, *The J Comparat Neurol* 1999, **408**:560-566.
23. **Zhao JH**, PC Sham, D Curtis. Letter to the Editor: A program for the Monte Carlo evaluation of significance of the extended TDT (ETDT). *Am J Hum Genet* 1999, **64**(5):1484-1485.
24. Curtis D, **JH Zhao**, PC Sham. Comparison of GENEHUNTER and MFLINK for analysis of COGA linkage data. *Genet Epidemiol* 1999, **17** (suppl 1):115-120.
25. **Zhao JH**, D Curtis, PC Sham. Model-free and permutation tests for allelic associations. *Hum Hered* 2000, **50**(2):133-139.
26. Li T, ZH Zhu, XH Liu, X Hu, **JH Zhao**, PC Sham, DA Collier. Association analysis of polymorphisms in the DRD4 gene and heroin in Chinese subjects. *Am J Med Genet* 2000, **96**:616-621.
27. Sham PC, MW Lin, **JH Zhao**, D Curtis. Power comparison of parametric and nonparametric linkage tests in small pedigrees. *Am J Hum Genet* 2000, **66**(5):1661-1668.
28. Sham PC, **JH Zhao**, D Curtis. The effect of marker polymorphism on the power to detect linkage disequilibrium due to single or multiple ancestral mutations. *Ann Hum Genet* 2000, **64**, 161-169.

□

29. Aitchison KJ, JG Frank, LC Quattrochi, A Sapone, **JH Zhao**, H Zaher, G Elizondo, C Bryant, JM, DA Collier, AJ Makoff, RW Kerwin. Identification of novel polymorphisms in the 5' flanking region of CYP1A2, characterization of interethnic variability, and investigation of their functional significance. *Pharmacogenet*, 2000, **10**:695-704.
30. Aitchison KJ, MW Jann, **JH Zhao**, T Sakai, H Zaher, K Wolff, AJ Makoff, DA Collier, RW Kerwin, FJ Gonzalez. Clozapine pharmacokinetics and Pharmacogenetics studied with CYP1A2-null mice. *J Psychopharmacol*, 2000, **14**, 353-359.
31. Li T, X Liu, Z Hong, **J Zhao**, X Hu, P Sham, D Collier. Association analysis of polymorphisms in the *mu* opiod gene and heroin abuse in Chinese subjects. *Addict Biol*, 2000, **5**:181-186.
32. Li T, X Liu, ZH Zhu, **J Zhao**, X Hu, DM Ball, PC Sham, DA Collier. No association between (AAT)_n repeats in the cannabinoid receptor gene (CNR1) and heroin abuse in a Chinese population. *Mol Psychiatr*, 2000 **5**, 128-130.
33. Li T, D Ball, **J Zhao**, RM Murray, X Liu, PC Sham, DA Collier. Family-based linkage disequilibrium mapping using SNP marker haplotypes: application to a potential locus for schizophrenia at chromosome 22q11. *Mol Psychiatr*, 2000, **5**, 77-84.
34. Sham P, **JH Zhao**, SS Cherny, JK Hewitt. Variance components QTL linkage analysis of selected and non-normal samples: conditioning on trait values. *Genet Epidemiol*, 2000, **19**, (suppl 1), 22-28
35. Sham PC, **JH Zhao**. The power of genome-wide sib pair linkage scans for quantitative trait loci using the new Haseman-Elston regression method, *GeneScreen* 2000, **1**:103-106.
36. Koch HG, J McClay, EW Loh, S Higuchi, **JH Zhao**, P Sham, D Ball and IW Craig. Allele association studies with SSR and SNP markers at known physical distances within a 1MB region embracing from the ALDH2 locus in the Japanese. *Hum Mol Genet*, 2000, **9**:2993-2999
37. Sham PC, **JH Zhao**, I Waldman, D Curtis. Should ambiguous trios for {TDT} be discarded? *Ann Hum Genet* 2000, **64**:575-576.
38. Karwautz A, S Rabe-Hesketh, X Hu, **J Zhao**, P Sham, DA Collier, JL Treasure. Individual-specific risk factors for anorexia nervosa: a pilot study using a discordant sister-pair design. *Psych Med* 2001, **31**(2):317-329.
39. Meira-Lima IV, **JH Zhao**, P Sham, AC Pereira, JE Krieger and H Vallada. Association and linkage studies between bipolar affective disorder and the polymorphic CAG/CTG repeat loci ERDA1, SEF2-1B, MAB21L and KCNN3, *Mol Psych* 2001, **6**(5):565-569.
40. Mill J, S Curran, L Kent, S Richards, A Gould, V Virdee, L Hockett, J Sharp, C Batten, S Fernando, E Simanoff, M Thompson, **J Zhao**, P Sham, E Taylor, P Asherson. Attention deficit hyperactivity disorder (ADHD) and the dopamine D4 receptor gene: evidence of association but no linkage in a UK sample. *Mol Psych* 2001, **6**(4): 440-444.
41. Cai G, T Li, H Deng, **J Zhao**, X Hu, RM Murray, X Liu, PC Sham, DA Collier. Affected sibling pair linkage analysis of qualitative and quantitative traits for schizophrenia on chromosome 22 in a Chinese population. *Am J Med Genet* 2001, **105**(4):321-327.
42. Russ C, JF Powerll, **J Zhao**, M Baker, M Hutton, F Crawford, M Mullan, G Roks, M Cruts, S Lovestone. The microtubule associated protein Tau gene and Alzheimer's disease - an association study and meta-analysis. *Neurosci Lett* 2001, **314**(1-2):92-96.
43. RYL Chen, P Sham, EYH Chen, T Li, EFC Cheung, TCK Hui, CL Kwok, F Lieh-Mak, **JH Zhao**, D Collier, R Murray. No association between T102C polymorphism of serotonin-2A receptor gene and clinical phenotypes of Chinese schizophrenic patients. *Psychitr Res* 2001, **105**: 175-185
44. **Zhao JH**, PC Sham. Faster allelic association using unrelated individuals. *Hum Hered* 2002, **53**: 36-41.
45. Li T, X Liu, **J Zhao**, X Hu, DM Ball, E-W Loh PC Sham and DA Collier. Allelic association analysis of the dopamine D2, D3, 5-HT(2A) and GABA(A)gamma2 receptors and the serotonin transporter genes with heroin abuse in Chinese subjects. *Am J Med Genet* 2002, **114**: 329-334.
46. Mallett R, J Leff, D Bhugra, D Pang, **JH Zhao**. Social environment, ethnicity and schizophrenia: a case-control study. *Social Psychiatr and Psychiatric Epidemiol* 2002, **37**: 329-335.

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47. **Zhao JH**, S Lissarrague, L Essioux, PC Sham. GENEcounting: haplotype analysis with missing genotypes. *Bioinformatics* 2002, **18**: 1694-1695.
48. **Zhao JH**, PC Sham. Generic number system and haplotype analysis. *Comp Meth Prog Biomed* 2003, **70**: 1-9.
49. Gabrovsek M, M Brecelj-Anderluh, L Bellodi, E Cellini, D Di Bella, X Estivill, F Fernandez-Aranda, B Freeman, F Geller, M Gratacos, R Haigh, J Hebebrand, A Hinney, J Holliday, X Hu, A Karwautz, B Nacmias, M Ribases, H Remschmidt, R Komel, S Sorbi, M Tomori, J Treasure, G Wagner, **J Zhao**, DA Collier. Combined family trio and case-control analysis of the COMT val158met polymorphism in European patients with anorexia nervosa *Am J Med Genet B (Neuropsychiatric Genet)* 2004, **124B**:68-72
50. Huang Y, T Li, Y Wang, J Ansar, G Lanting, X Liu, **JH Zhao**, X Hu, PC Sham, D Collier. Linkage disequilibrium analysis of polymorphisms in the gene for myelin oligodendrocyte glycoprotein in Tourette's syndrome patients from a Chinese sample. *Am J Med Genet. (Neuropsychiatric Genet)* 2004, **124B**:76-80.
51. Shi J, S Zhang, C Ma, X Liu, T Li, M Tang, H Han, Y Guo, **JH Zhao**, K Zheng, X Kong, K Zhang, Z Su, Z Zhao. Association between apolipoprotein C1 HpaI polymorphism and sporadic Alzheimer's disease in Chinese. *Acta Neurol Scan* 2004, **109**:140-145.
52. Shi J, S Zhang, M Tang, X Liu, T Li, H Han, Y Wang, Y Guo, **J Zhao**, H Li, C Ma. Possible association between Cys311Ser polymorphism of paraoxonase 2 gene and late-onset Alzheimer's disease in Chinese. *Mol Brain Res* 2004, **120**:201-204.
53. Tan Q, **JH Zhao**, I Iachine, J Hjelmberg, W Vach, JW Vaupel, Christensen K, TA Kruse. Power of non-parametric linkage analysis in mapping genes contributing to human longevity *Genet Epidemiol* 2004, **26**:245-253.
54. **Zhao JH**. 2LD, GENEcounting and HAP: Computer programs for linkage disequilibrium analysis. *Bioinformatics* 2004, **20**:1325-1326.
55. **Zhao JH**, Book review: Lachin JM (2000): Biostatistical methods: the assessment of relative risks. New York: John Wiley. *Stat Methods Med Res* 2004; **13**: 414-415.
56. Parsian A, R Sinha, B Racette, **JH Zhao**, JS Perlmutter. Association of a variation in the promoter of the brain-derived neurotrophic factor gene with familial parkinson's disease. *Parkinsonism and Related Disorders* 2004, **10**:213-219.
57. Walshe M, C McDonald, M Taylor, **J Zhao**, P Sham, A Grech, K Schulze, E Bramon, R Murray. Obstetric complications in patients with Schizophrenia and their unaffected siblings *European Psychiatr* 2005, **20**:28-34.
58. Shi J, S Zhang, M Tang, C Ma, **J Zhao**, T Li, X Liu, Y Sun, Y Guo, H Han, Y Ma, Z Zhao. Mutation screening and association study of the neprilysin gene in sporadic Alzheimer's disease in Chinese persons. *J Gerontology Bio Sci* 2005, **60A**: 301-306.
59. Tan Q, L Christiansen, L Bathum, **JH Zhao**, AI Yashin, JW Vaupel, K Christensen, TA Kruse. Estimating haplotype relative risks on human survival in population-based association studies. *Hum Hered* 2005, **59**:88-97.
60. Tan Q, L Christiansen, L Bathum, **JH Zhao**, W Vach, JW Vaupel, K Christensen, TA Kruse. Haplotype effects on human survival: logistic regression models applied to unphased genotype data. *Ann Hum Genet* 2005, **69**: 168-175.
61. **Zhao JH**, EJ Brunner, M Kumari, A Singh-Manoux, E Hawe, PJ Talmud, MG Marmot, SE Humphries. APOE polymorphism, socioeconomic status and cognitive function in later mid-life: The Whitehall II longitudinal study. *Soc Psychiatr and Psychiatr Epidemiol* 2005, **40**:557-563.
62. **Zhao JH**. Mixed-effects Cox models of alcohol dependence in extended families. *BMC Genetics* 2005, (Suppl) **6**:127.
63. Tan Q, K Christensen, L Christiansen, L Bathum, S Li, **JH Zhao**, TK Kruse. Haplotype association analysis of human disease traits using multi-locus genotype data of unrelated subjects. *Genetical Res* 2005, **86**: 223-231.

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64. **Zhao JH**, Q Tan. Integrated analysis of genetic data with R. *Hum Genomics* 2006, **2**(4):258-265.
65. **Zhao JH**, Drawing pedigree diagrams with R and graphviz, *R News* 6:38-41, 2006
66. **Zhao JH**. Pedigree-drawing with R and graphviz. *Bioinformatics* **22**(8):1013-1014.
67. **Zhao JH**, Q Tan. Genetic dissection of complex traits *in silico*: approaches, problems and solutions. *Curr Bioinformatics* 2006, **1**:359-369.
68. **Zhao JH**, Luan JA, Tan Q, Loos R, Wareham NJ. Analysis of large genomic data *in silico*: the EPIC-Norfolk study of obesity. In DS Huang, L Heutte, and M Loog (Eds). *Advanced Intelligent Computing Theories and Applications with Aspects of Contemporary Intelligent Computing Techniques*, Third International Conference on Intelligent Computing (ICIC) 2007: 781-790.
69. Parsian AJ, Racette BA, **Zhao JH**, Sinha R, Patra B, Perlmutter JS, Parsian A. Association of alpha-synuclein gene haplotypes with Parkinson's disease. *Parkinsonism Relat Disord* 2007 Aug;**13**(6):343-347.
70. Tan Q, Christiansen L, Brasch-Andersen C, **Zhao JH**, Kruse TA, Christensen K. Retrospective analysis of main and interaction effects in genetic association studies of human complex traits. *BMC Genet* 2007, **8**:70.
71. **Zhao JH**, J Luan, F Baksh, Q Tan. Mining gene networks with application to GAW15 problem 1. *BMC Proc* 2007, **1** (Suppl 1):S52
72. **Zhao JH**. gap: genetic analysis package. *J Stat Soft* 2007, **23** (8):1-18.
73. Sandhu MS, et al. LDL-cholesterol concentrations: a genome-wide association study. *Lancet* 2008, **371**:483-491.
74. Tan Q, M Thomassen, KM Jochumsen, **JH Zhao**, K Christensen, TA Kruse. Evolutionary Algorithm for Feature Subset Selection in Predicting Tumor Outcomes Using Microarray Data. I. Măndoiu, R. Sunderraman, and A. Zelikovsky (Eds.): *ISBRA 2008*, LNBI 4983, pp. 426–433, 2008. © Springer-Verlag Berlin Heidelberg 2008
75. Weedon MN, et al. Genome-wide association analysis identifies 20 loci that influence adult height. *Nat Genet* 2008, **40**:575-583.
76. Loos R, et al. Common variants near *MC4R* are associated with fat mass, weight and risk of obesity. *Nat Genet* 2008, **40**:768-775.
77. Tan Q, **J Zhao**, S Li, L Christiansen, TA Kruse, K Christensen. Differential and correlation analyses of microarray gene expression data in the CEPH Utah families. *Genomics* 2008, **92**:94-100.
78. Tan Q, **JH Zhao**, TA Kruse, K Christensen. Power for genetic association study of human longevity using the case-control design. *Am J Epidemiol* 2008, **168**:890-896.
79. E Vassos, PC Sham, G Cai, H Deng, X Liu, X Sun, **J Zhao**, RM Murray, DA Collier, T Li. Correlation and familial aggregation of dimensions of psychosis in affected sibling pairs from China. *Brit J Psychiat* 2008, **193**:305-310.
80. Barroso I, JA Luan, E Wheeler, P Whittaker, J Wasson, E Zeggini, MN Weedon, S Hunt, R Venkatesh, TM Frayling, M Delgado, RJ Neuman, **J Zhao**, R Sherva, B Glaser, M Walker, G Hitman, MI McCarthy, AT Hattersley, MA Permutt, NJ Wareham, P Deloukas. Population-specific risk of type 2 diabetes (T2D) conferred by *HNF4A* P2 promoter variants: a lesson for replication studies. *Diabetes* 2008, **57**:3161-3165.
81. Willer CJ, et al. Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. *Nat Genet*, 2009, **41**:25-34.
82. Prokopenko I, et al. Variants in *MTNR1B* influence fasting glucose levels. *Nat Genet*, 2009, **41**:77-81.
83. Patra B, Parsian AJ, Racette BA, **Zhao JH**, Perlmutter JS, Parsian A. LRRK2 gene G2019S mutation and SNPs [haplotypes] in subtypes of Parkinson's disease. *Parkinsonism Relat Disord*, 2009, **15**:175-180.
84. Ong KK, Elks CE, Li S, **Zhao JH**, Luan J, Andersen LB, Bingham SA, Brage S, Smith GD, Ekelund U, Gillson CJ, Glaser B, Golding J, Hardy R, Khaw KT, Kuh D, Luben R, Marcus M,



- McGeehin MA, Ness AR, Northstone K, Ring SM, Rubin C, Sims MA, Song K, Strachan DP, Vollenweider P, Waeber G, Waterworth DM, Wong A, Deloukas P, Barroso I, Mooser V, Loos RJ, Wareham NJ. Genetic variation in *LIN28B* is associated with the timing of puberty. *Nat Genet* 2009, **41**:729-733.
85. Newton-Cheh C, et al. Genome-wide association study identifies eight loci associated with blood pressure. *Nat Genet* 2009, **41**:666-676.
86. Lindgren CM, et al. Genome wide association scan meta-analysis identifies three loci influencing adiposity and fat distribution, *PLoS Genetics*. 2009, **5**(6):e1000508.
87. Vimalaswaran KS, Li S, **Zhao JH**, Luan JA, Bingham SA, Khaw K-T, Ekelund U, Wareham NJ, Loos RJF. Physical activity attenuates the BMI-increasing influence of genetic variation in *FTO*. *Am J Clin Nutr* 2009, **90**:425-428.
88. Beardsal K, Ong KK, Murphy N, Ahmed ML, **Zhao JH**, Peeters M, Dunger DB. Heritability of childhood weight gain from birth and risk markers for adult disease in prepubertal twins. *JCEM*. 2009, **94**:3708-3713.
89. Luan J, Kerner B, **Zhao JH**, Loos RJ, Sharp SJ, Muthen BO, Wareham NJ. A multilevel linear mixed model of the association between candidate genes and weight and body mass index using the Framingham longitudinal family data, *BMC Proc*. 2009, **3**(Suppl 7):S115.
90. Li S, **Zhao JH**, Luan J, Luben RN, Rodwell SA, Khaw K-T, Ong KK, Wareham NJ, Loos RJF. Cumulative effects and predictive value of common obesity-susceptibility variants identified by genome-wide association studies. *Am J Clin Nutr*. 2010, **91**:184-190.
91. Zhao, J. H. Genetic association analysis with R. *The Biomedical & Life Sciences Collection* **2009**, e1002430 (2009), <https://doi.org/10.69645/DCRY5578>.
92. Repapi E, et al. Genome-wide association study identifies five loci associated with lung function. *Nat Genet*. 2010, **42**(1):36-44.
93. Dupuis J, et al. Novel genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. *Nat Genet*. 2010, **42**:105-116
94. Saxena R, et al. Genetic variation in *GIPR* influences the glucose and insulin responses to an oral glucose challenge. *Nat Genet* 2010, **42**:142-148
95. Peng Q, **Zhao JH**, Xue F. PCA-based bootstrap confidence interval tests for gene-disease association involving multiple SNPs. *BMC Genet* 2010, **11**:6
96. Peng Q, **Zhao JH**, Xue F. A gene-based method for detecting gene-gene co-association in a case-control association study. *Eur J Hum Genet* 2010, **18**:582-587
97. Liu JZ, et al. Meta-analysis and imputation refines the association of 15q25 with smoking quantity. *Nat Genet* 2010, **42**(5):436-40
98. Teslovich et al. Biological, clinical, and population relevance of 95 loci mapped for serum lipid concentrations. *Nature* 2010, **466**:707-13
99. Lango Allen, H., K. Estrada, et al. Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature* 2010, **467**(7317): 832-838.
100. Tan Q, **Zhao JH**, Li S, Kruse TA, Christensen K. Power assessment for genetic association study of human longevity using offspring of long-lived families. *Eur J Epidemiol* **21**:501-506, 2010.
101. Yang Q, et al. Racial/Ethnic Differences in Association of Fasting Glucose–Associated Genomic Loci With Fasting Glucose, HOMA-B, and Impaired Fasting Glucose in the U.S. Adult population *Diabetes Care* 2010, **33**:2370-2377.
102. den Hoed M, Ekelund U, Brage S, Grontved A, **Zhao JH**, Sharp SJ, Ong KK, Wareham NJ, Loos RJ. Genetic Susceptibility to Obesity and Related Traits in Childhood and Adolescence: Influence of Loci Identified by Genome-Wide Association Studies. *Diabetes* 2010, **59**:2980-2988.
103. Speliotes EK, et al. Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nat Genet* 2010, **42**(11):937-948.
104. Heid IM, et al. Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution *Nat Genet* 2010, **42**(11):949-960.

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105. Elks CE, et al. Thirty new loci for age at menarche identified by a meta-analysis of genome-wide association studies *Nat Genet* 2010; **42**:1077-1085
106. Vimalaswaran KS, **Zhao JH**, Wainwright NW, Surtees PG, Wareham NJ, Loos RJ. Association between serotonin 5-HT-2C receptor gene (HTR2C) polymorphisms and obesity- and mental health-related phenotypes in a large population-based cohort. *Int J Obe (Lond)*. 2010; **34**(6):1028-33.
107. Li S, **Zhao JH**, Luan JA, Ekelund U, Luben RN, Khaw K-T, Wareham NJ, Loos RJF. Physical activity attenuates the genetic predisposition to obesity in 20,000 men and women of European descent. *PLoS Med* 2010; **7**(8): e1000332
108. Waterworth DM, et al. Genetic Variants Influencing Circulating Lipid Levels and Risk of Coronary Artery Disease. *Arterioscler Throm Vasc Biol*, 2010; **30**:2264-2276
109. Bhugra D, Leff J, Mallett R, Morgan C, **Zhao JH**. The culture and identity schedule a measure of cultural affiliation: acculturation, marginalization and schizophrenia. *Int J Soc Psychiatr* 2010, **56**(5):540-56
110. Böger CA, et al. CUBN is a gene locus for albuminuria. *JASN* 2011, **22**:555-570.
111. Schumann G, et al. Genome-wide association and genetic functional studies identify AUTS2 in the regulation of alcohol consumption. *PNAS* 2011; **108**:7119-7124
112. Obeidat M, et al. A Comprehensive evaluation of potential lung function associated genes in SpiroMeta general population sample. *PLoS One* 2011; **6**:e19382
113. **Zhao JH**, Luan JA, Loos RJF, Wareham N. On genotype-phenotype association using SAS. *Proc IASTED Int Conf Comp Bios (CompBio 2011)* 742-040:428-433
114. Kilpeläinen TO, et al. Genetic variation near IRS1 associates with reduced adiposity and an impaired metabolic profile. *Nat Genet* 2011;**43**(8):753-866
115. Gao Q, He Y, Yuan Z, **Zhao J**, Zhang B, Xue F. Gene- or region-based association study via kernel principal component analysis. *BMC Genet* 2011; **12**:75
116. Soler Artigas M, et al. *Am J Respir Crit Care Med* 2011; **184**:785-795
117. Soler Artigas M, et al. Genome-wide association and large-scale follow up identifies 16 new loci influencing lung function. *Nat Genet*. 2011; **43**(11):1082-90
118. Wain LV, et al. Genome-wide association study identifies six new loci influencing pulse pressure and mean arterial pressure. *Nat Genet* 2011; **43**:1005-1012
119. International Consortium for Blood Pressure Genome-Wide Association Studies, Ehret GB, et al. Genetic variants in novel pathways influence blood pressure and cardiovascular disease risk. *Nature* 2011; **478**:103-109
120. Gieger C, et al. New gene functions in megakaryopoiesis and platelet formation. *Nature* 2011; **480**:201-208
121. Li S, Zhao JH, Luan J, Langenberg C, Luben RN, Khaw KT, Wareham NJ, Loos RJ. Genetic predisposition to obesity leads to increased risk of type 2 diabetes. *Diabetologia*. 2011;**54**(4):776-82.
122. Elks CE, den Hoed M, **Zhao JH**, Sharp SJ, Wareham NJ, Loos RJF, Ong KK. Variability in the heritability of body mass index: a systematic review and meta-regression. *Front Endocrinol* 2012; **3**(29):1-16
123. Xue F, Li S, Luan J, Yuan Z, Luben RN, Khaw K-T, Wareham NJ, Loos RJF, **Zhao JH**. A latent variable partial least squares path modeling approach to regional association and polygenic effect with applications to a human obesity study. *PLoS ONE* 2012; **7**(2): e31927
124. **Zhao JH**, Luan JA. Mixed modeling with whole genome data. *J Prob Stat*. 2012. doi 10.1155/2012.485174
125. Yang J, et al. Genetic effects on variability: *FTO* genotype is associated with phenotypic variance of body mass index. *Nature* 2012;**490**(7419):267-72

□

126. Vimalleswaran KS, Tachmazidou I, **Zhao JH**, Hirschhorn JN, Dudbridge F, Loos RJF. Candidate genes for obesity-susceptibility show enriched association within a large genome-wide association study for BMI. *Hum Mol Genet* 2012;21(20):4537-42
127. Boraska V, et al. Genome-wide meta-analysis of common variant differences between men and women. *Hum Mol Genet* 2012; 21(21): 4805-4815
128. Wilk JB, et al. Genome-Wide Association Studies Identify CHRNA5/3 and HTR4 in the Development of Airflow Obstruction. *Am J Respir Crit Care Med* 2012;186(7):622-32
129. Ramasamy A., et al. Genome-wide association studies of asthma in population-based cohorts confirm known and suggested loci and identify an additional association near HLA. *PLoS One* 7(9): e44008. doi:10.1371/journal.pone.0044008
130. Yuan Z, Gao Q, He Y, Zhang X, Li F, **Zhao J**, Xue F. Detection for gene-gene co-association via kernel canonical correlation analysis. *BMC Genet* 2012; 13:83
131. Hancock DB, et al. Genome-wide joint meta-analysis of SNP and SNP-by-smoking interaction identifies novel loci for pulmonary function. *PLoS Genet* 2012; 8(12): e1003098
132. Van der Harst P, et al. 75 genetic loci influencing the human red blood cell. *Nature* 2012; 492:369-375
133. Morris AP, et al. Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. *Nat Genet* 2012; 44:981-990
134. Horikoshi M, et al. Novel loci associated with birth weight reveal genetic links between intrauterine growth and adult height and metabolism. *Nat Genet* 2013; 45:76-82
135. Kottgen A., et al. Genome-wide association analyses identify 18 new loci associated with serum urate concentrations. *Nat Genet* 2013; 45:145-154
136. Berndt SL, et al. Large-scale genome-wide meta-analysis identifies 11 novel loci for anthropometric traits and provides new insights on the genetic architecture of the extremes of the distribution. *Nat Genet* 2013;45:501-512
137. den Hoed M, et al. Heart rate-associated loci and their effects on cardiac conduction and rhythm disorders. *Nat Genet* 2013;45:621-631
138. Monda KL, et al. A meta-analysis identifies new loci associated with body mass index in individuals of African ancestry. *Nat Genet* 2013; 45:690-696
139. Randall JC, et al. Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. *PLoS Genet* 2013; 9(6): e1003500
140. Zhang X, Yang X, Yuan Z, Liu Y, Li F, Peng B; Zhu D, Zhao J, Xue F. A PLSPM-based test statistic for detecting gene-gene co-association in genome-wide association study with case-control design. *PLoS One* 2013; 8(4):e62129. doi: 10.1371/journal.pone.0062129
141. Yuan Z, Liu H, Zhang X, Li F, Zhao J, Zhang F, Xue F. From interaction to co-association-A Fisher r-to-z transformation-based novel statistic for real world genome-wide association study. *PLoS One* 2013; 8(7): e70774
142. van Vliet-Ostaptchouk JV, et al. Pleiotropic effects of obesity-susceptibility loci on metabolic traits: a meta-analysis of up to 37,874 individuals. *Diabetologia* 2013; 56:2134–2146
143. Li F, Zhao J, Yuan Z, Zhang X, Ji J, Xue F. A powerful latent variable method for detecting and characterizing gene-based gene-gene interaction on multiple quantitative traits. *BMC Genet* 2013, 14:89 doi:10.1186/1471-2156-14-89.
144. Yaghootkar H, et al. Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and Type 2 diabetes. *Diabetes* 2013; 62:3589-3598
145. den Hoed M, Brage S, Zhao JH, Westgate K, Nessa A, Ekelund U, Spector TD, Wareham NJ, Loos RJF. Heritability of objectively assessed daily physical activity. *Am J Clin Nutr* 2013; 98:1317-1325



146. Willer CJ, et al. Discovery and refinement of loci associated with lipid levels. *Nat Genet* 2013; 45:1274-1283
147. Do R, et al. Common variants associated with plasma triglycerides and risk for coronary artery disease. *Nat Genet* 2013; 45:1345-1352
148. Cheng CY, et al. Nine loci for ocular axial length identified through genome-wide association studies, including shared loci with refractive error. *Am J Hum Genet.* 2013 Aug 8;93(2):264-77. doi: 10.1016/j.ajhg.2013.06.016. PMID: 24144296; PMCID: PMC3772747.
149. Tan Q, Hjelmberg JvB, Thomassen M, Jensen AK, Christiansen L, Christensen K, Zhao J, Kruse TA. Hierarchical linear modelling of longitudinal pedigree data for genetic association analysis. *BMC Proc* 2014, 8(Suppl 1):S82, doi:10.1186/1753-6561-8-S1-S82
150. Loth DW, et al. Genome-wide association analysis identifies six new loci associated with forced vital capacity. *Nat Genet* 2014, 46(7):669-77
151. Simino J, et al. Gene-Age Interactions in Blood Pressure Regulation: A Large-Scale Investigation with the CHARGE, Global BPgen, and ICBP Consortia. *Am J Hum Genet* 2014, 95: 24–38
152. Hoggart CJ, et al. Novel approach identifies SNPs in *SLC2A10* and *KCNK9* with evidence for parent-of origin effect on body mass index. *PLoS Genet* 2014, 10(7): e1004508
153. Tan Q, Zhao JH, Kruse T, Christensen K. Power estimation for gene-longevity association analysis using concordant twins. *Genet Res Int* 2014, Article ID 154204, <http://dx.doi.org/10.1155/2014/154204>
154. Perry JRB, et al. Parent-of-origin specific allelic associations among 106 genomic loci for age at menarche. *Nature* 2014, 514:92-97
155. Wood AR. Defining the role of common variation in the genomic and biological architecture of adult human height. *Nat Genet* 2014, 46: 1173–1186
156. Wessel, J, et al. Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. *Nat Comm* 2015, 6:5897, DOI: 10.1038/ncomms689
157. Cornelis MC, et al. Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. *Mol Psychiatr* 2015, 20(5); 647-656
158. Shungin D, et al., New genetic loci link adipose and insulin biology to body fat distribution. *Nature* 2015, 518:187-196
159. Locke AE, et al. Large-scale genetic studies of body mass index provide insight into the biological basis of obesity. *Nature* 2015, 518,197–206
160. Zimmermann E, et al. Is the adiposity-associated *FTO* gene variant related to all-cause mortality independent of adiposity? Meta-analysis of data from 169,551 white Caucasian adults. *Obesity Rev* 2015, 16:327-340
161. Joshi PK, et al. Directional dominance on stature and cognition in diverse human populations. *Nature* 2015, 523:459-462
162. Gharib S, et al. Integrated pathway genomics of lung function and airflow obstruction. *Hum Mol Genet* 2015 24(23):6836–6848
163. Winkler TW, et al. The influence of age and sex on genetic associations with adult body size and shape: A large-scale genome-wide interaction study. *PLoS Genet* 2015, DOI: 10.1371/journal.pgen.1005378
164. Obeidat M, et al. Molecular mechanisms underlying variations in lung function: a systems genetics analysis. *Lancet Res Med* 2015, DOI: [http://dx.doi.org/10.1016/S2213-2600\(15\)00380-X](http://dx.doi.org/10.1016/S2213-2600(15)00380-X)
165. Solar Artigas M, et al. Sixteen new lung function signals identified through 1000 Genomes Project reference panel imputation. *Nat Comm* 2015, 6, Article number: 8658 doi:10.1038/ncomms9658

□

166. Fan Q, et al. Meta-analysis of gene–environment-wide association scans accounting for education level identifies additional loci for refractive error. *Nat Comm* 2015, 7:11008 | doi: 10.1038/ncomms11008
167. de Vries PS, et al. A meta-analysis of 120,246 individuals identifies 18 new loci for fibrinogen concentration. *Hum Mol Genet* 2016, 25(2): 368-370
168. Lu Y, et al. New loci for body fat percentage reveal link between adiposity and cardiometabolic disease risk. *Nat Comm* 2016, 7. 10495, doi:10.1038/ncomms10495
169. Kilpeläinen TO, et al. Genome-wide meta-analysis uncovers novel loci influencing circulating leptin levels. *Nat Comm* 2016, 7,10494, doi:10.1038/ncomms10494
170. Teumer A, et al. Genome-wide Association Studies Identify Genetic Loci Associated with Albuminuria in Diabetes *Diabetes* 2016;65:803-817
171. Wang S., et al. General framework for meta-analysis of haplotype association tests. *Genet Epidemiol* 2016; 40(3):244-252
172. Scott RA, et al. A genomic approach to therapeutic target validation identifies a glucose-lowering GLP1R variant protective for coronary heart disease. *Sci Trans Med* 2016; 8: 341ra76
173. Horikoshi M, et al. Genome-wide associations for birth weight and correlations with adult diseases. *Nature* 2016; 538:248-252
174. Ehret GB, et al. The genomics of blood pressure regulation and its target organs from association studies in 342,415 individuals *Nat Genet* 2016;48(10):1171-84
175. Barban B, et al. Genome-wide analysis identifies 12 loci influencing human reproductive behaviour. *Nat Genet* 2016; 48(12):1462-1472
176. Schumann G, et al. *KLB* is associated with alcohol drinking, and its gene product β -Klotho is necessary for *FGF21* regulation of alcohol preference. *PNAS* 2016; 113: 14372–14377
177. Ried JS, et al. A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. *Nat Comm* 2016; doi: 10.1038/ncomms13357
178. Aschard H, et al. Evidence for large-scale gene-by-smoking interaction effects on pulmonary function. *Int J Epidemiol* 2017;46(3):894-904. doi: 10.1093/ije/dyw318
179. Marouli E, et al. Rare and low-frequency coding variants alter human adult height. *Nature* 2017; 542(7640):186-190. doi: 10.1038/nature21039
180. Wain LV, et al. Genome-wide association analyses for lung function and chronic obstructive pulmonary disease identify new loci and potential druggable targets. *Nat Genet* 2017; 49(3):416-425. doi: 10.1038/ng.3787
181. Justice AE, et al. Genome-wide meta-analysis of 241,258 adults accounting for smoking behavior identifies novel loci for obesity traits. *Nat Comm* 2017; 8:14977. doi: 10.1038/ncomms14977
182. Day F, et al. Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. *Nat Genet* 2017; 49(6):834-841. doi: 10.1038/ng.3841
183. Mullin BH, et al. Genome-wide association study meta-analysis for quantitative ultrasound parameters of bone identifies five novel loci for broadband ultrasound attenuation. *Hum Mol Genet* 2017; 26(14):2791-2802. doi: 10.1093/hmg/ddx174
184. Zillikens CM, et al. A large meta-analysis of genome wide association studies identifies five significant loci for lean body mass. *Nat Comm.* 2017; 8(1):80. doi: 10.1038/s41467-017-00031-7.
185. Graff M, et al. Genome-wide physical activity interactions in adiposity - a meta-analysis of 200,452 adults. *PLoS Genet* 2017; 13(8):e1006972. doi: 10.1371/journal.pgen.1006972.
186. Saleheen D, et al. Loss of cardio-protective effects at the *ADAMTS7* locus due to gene-smoking interactions. *Circulation* 2017; 135(24):2336-2353
187. Wain L, et al. Novel blood pressure locus and gene discovery using genome-wide association and expression data sets from blood and the kidney. *Hypertension* 2017 70:e4–e19, doi: 10.1161/HYPERTENSIONAHA.117.09438.

□

188. Demenais F, et al. Multi-ancestry genome-wide association study identifies new asthma susceptibility loci that co-localize with immune cell enhancer histone marks. *Nat Genet* 2018; 50(1):42-53
189. Medina-Gomez C, et al. Life-course genome-wide association study meta-analysis of total body BMD and assessment of age-specific effects. *Am J Hum Genet* 2018; 102(1):88-102
190. Sung YJ, et al. A large-scale multi-ancestry genome-wide study accounting for smoking behavior identifies multiple significant loci for blood pressure. *Am J Hum Genet* 2018, 102(3):375-400. doi: 10.1016/j.ajhg.2018.01.015.
191. Turcot V, et al. (2018). Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure underpinning obesity. *Nat Genet* 50:26-41
192. **Zhao JH**, Luan JA, Congdon P. Bayesian linear mixed model of polygenic effects. *J Stat Soft* 2018, 85(6):1-27. doi: 10.18637/jss.v085.i06
193. Feitosa MF, et al. Novel genetic associations for blood pressure identified via gene-alcohol interaction in up to 570K individuals across multiple ancestries. *PLoS One* 2018, 13(6):e0198166. doi: 10.1371/journal.pone.0198166.
194. Lee JJ, et al. Gene discovery and polygenic prediction from a genome-wide association study of educational attainment in 1.1 million individuals. *Nat Genet* 2018, 50:1112–1121, <https://doi.org/10.1038/s41588-018-0147-3>.
195. Ligthart S, et al. Genome analyses of >200,000 individuals identify 58 loci for chronic inflammation and highlight pathways that link inflammation and complex disorders. *Am J Hum Genet* 2018, 103(5):691-706. doi: 10.1016/j.ajhg.2018.09.009.
196. Evangelou E, et al. Genetic analysis of over 1 million people identifies 535 new loci for blood pressure traits. *Nat Genet* 2018, 50(10):1412-1425. doi: 10.1038/s41588-018-0205-x.
197. Merino J, et al. Genome-wide meta-analysis of macronutrient intake of 91,114 European ancestry participants from the cohorts for heart and aging research in genomic epidemiology consortium. *Mol Psychiatr* 2018 Jul 9. doi: 10.1038/s41380-018-0079-4.
198. Kilpeläinen TO, et al. Multi-ancestry study of blood lipid levels identifies four loci interacting with physical activity. *Nat Comm* 2019, 10, 376, <https://doi.org/10.1038/s41467-018-08008-w>.
199. Giri A, et al. Trans-ethnic association study of blood pressure determinants in over 750,000 individuals. *Nat Genet* 2019, 51:51-62
200. Karasik D, et al. Disentangling the genetics of lean mass. *Am J Clin Nutr* 2019, 109(2): 276-287, <https://doi.org/10.1093/ajcn/nqy272>.
201. de Vries PS, et al. Multi-ancestry genome-wide association study of lipid levels incorporating gene-alcohol interactions. *Am J Epidemiol* 2019, 188(6):1033-1054, <https://doi.org/10.1093/aje/kwz005>.
202. Justice AE, et al. Protein-coding variants highlight the importance of lipolysis in adipocytes for body fat distribution. *Nat Genet* 2019, <https://doi.org/10.1038/s41588-018-0334-2>.
203. Shrine N, et al. New genetic signals for lung function highlight pathways and chronic obstructive pulmonary disease associations across multiple ancestries, *Nat Genet* 2019, <https://doi.org/10.1038/s41588-018-0321-7>.
204. Zhao J, et al. Meta-analysis of genome-wide association studies provides insights into genetic control of tomato flavor. *Nat Comm* 2019, 10, Article number: 1534 (2019), <https://www.nature.com/articles/s41467-019-09462-w>
205. Sung YJ, et al. A multi-ancestry genome-wide study incorporating gene–smoking interactions identifies multiple new loci for pulse pressure and mean arterial pressure. *Hum Mol Genet* 2019, doi: 10.1093/hmg/ddz070
206. Clark DW, et al. Associations of autozygosity with a broad range of human phenotypes. *Nat Comm* 2019, NCOMMS-18-33232A-Z

□

207. Bentley A.R., *et al.* Multi-ancestry genome-wide gene–smoking interaction study of 387,272 individuals identifies new loci associated with serum lipids. *Nat Genet* **51**, 636–648 (2019). <https://doi.org/10.1038/s41588-019-0378-y>
208. Shah S, et al. Genome-wide association and Mendelian randomisation analysis provide insights into the pathogenesis of heart failure. *Nat Comm* 2020, 11(1):163. Published 2020 Jan 9. doi:10.1038/s41467-019-13690-5
209. Surendran P, et al. Discovery of rare variants associated with blood pressure regulation through meta-analysis of 1.3 million individuals. *Nat Genet* 2020, 52(12):1314–1332, doi: 10.1038/s41588-020-00713-x.
210. Lin W, Ji J, Zhu Y, Li M, Zhao J, Xue F, Yuan Z. PMINR: pointwise mutual information-based network regression – with application to studies of lung cancer and Alzheimer’s disease. *Front Genet* 2020, 11:556259, <https://doi.org/10.3389/fgene.2020.556259>
211. Cuellar-Partide G, et al. Genome-wide association study identifies 48 common genetic variants associated with handedness. *Nat Hum Behaviour* 2021, 5: 59–70.
212. Gaziano L, et al. Actionable druggable genome-wide Mendelian randomization identifies repurposing opportunities for COVID-19. *Nat Med* 2021, 27:668–676.
213. Chen J, et al. The trans-ancestral genomic architecture of glycemic traits. *Nat Genet* 2021, 53:840–860.
214. COVID-19 Host Genetics Initiative. Mapping the human genetic architecture of COVID-19. *Nature* 2021.
215. Zhang Y, et al. Mendelian randomisation highlights hypothyroidism as a causal determinant of idiopathic pulmonary fibrosis. *EBiomed.* 2021, <https://doi.org/10.1016/j.ebiom.2021.103669>
216. Graham SE, et al. The power of genetically diverse individuals in genome-wide association studies of blood lipid levels. *Nature* 2021, **600**: 675–679, DOI:10.1038/s41586-021-04064-3
217. Jin X, Zhang L, Ji J, Ju T, Zhao JH, Yuan Z. NeRiT -- Network regression in transcriptome-wide association studies. *BMC Genomics* (2022) 23:562, <https://doi.org/10.1186/s12864-022-08809-w>
218. Pathak, G.A. *et al.* A first update on mapping the human genetic architecture of COVID-19. *Nature* **608**, E1–E10 (2022).
219. Ramdas S, et al. A multi-layer functional genomic analysis to understand noncoding genetic variation in lipids. *Am J Hum Genet.* 2022 Aug 4;**109**(8):1366–1387. doi: 10.1016/j.ajhg.2022.06.012.
220. Wang Z, et al. Genome-wide association analyses of physical activity and sedentary behavior provide insights into underlying mechanisms and roles in disease prevention *Nat Genet.* 2022, **54**:1332–1344
221. Yengo L, et al. A saturated map of common genetic variants associated with human height from 5.4 million individuals of diverse ancestries. *Nature* 2022, **610**:704–712.
222. Kanoni, S., et al. Implicating genes, pleiotropy, and sexual dimorphism at blood lipid loci through multi-ancestry meta-analysis. *Genome Biology* 2022, **23**(1): 268 (medRxiv, DOI:10.1101/2021.12.15.21267852).
223. Shrine N, et al. Multi-ancestry genome-wide association study improves resolution of genes, pathways and pleiotropy for lung function and chronic obstructive pulmonary disease. *Nat Genet* 2023, **55**:410–422
224. van de Vegte JY, et al. Genetic insights into resting heart rate and its role in cardiovascular disease. *Nat Comm* **14**:4646 (2023), <https://www.nature.com/articles/s41467-023-39521-2>
225. **Zhao JH**, et al. Mapping pQTLs of circulating inflammatory proteins identifies drivers of immune-mediated disease risk and novel therapeutic targets. *Nat Immunol* 2023, **24**(9):1540–1551, 10.1038/s41590-023-01588-w, <https://www.nature.com/articles/s41590-023-01588-w>.
226. Lagou V, et al. GWAS of random glucose in 476,326 individuals provide insights into diabetes pathophysiology, complications and treatment stratification. *Nat Genet* **55**:1448–1461 (2023),



<https://www.nature.com/articles/s41588-023-01462-3>, *medRxiv*, DOI: 10.1101/2021.04.17.21255471.

227. The Host Genetics Initiative. A second update on mapping the human genetic architecture of COVID-19. *Nature* **621**:E7–E26 (2023).
228. Macdonald-Dunlop E, et al. Mapping genetic determinants of 184 circulating proteins in 26,494 individuals to connect proteins and diseases. *medRxiv*, DOI:10.1101/2021.08.03.21261494.
229. Klaric L, et al. Mendelian randomisation identifies alternative splicing of the FAS death receptor as a mediator of severe COVID-19. *medRxiv*, doi: <https://doi.org/10.1101/2021.04.01.21254789>.
230. Gaziano L, et al. Transcriptome- and proteome-wide Mendelian randomization to prioritize therapeutic targets for coronary heart disease. *medRxiv*, DOI: 10.1101/2024.06.27.24309406v1.
231. Zhao JH. Genetic association analysis with R (II). *The Biomedical & Life Sciences Collection* **2025**, <https://github.com/jinghuazhao/gaawr2>, (2025).
232. Kelemen M, et al. Performance of deep-learning based approaches to improve polygenic scores. *Nat Comm* **16**, 5122 (2025). <https://doi.org/10.1038/s41467-025-60056-1>.
233. Smit RAJ, et al. Polygenic scores to predict body mass index and obesity across populations and through the life course powered by data from 5.1 million individuals, *Nat Med* (2025). <https://www.nature.com/articles/s41591-025-03827-z>.
234. Koprulu M, et al. Large-scale proteogenomic meta-analyses highlight N-linked glycosylation as important regulator of the circulating proteome.
235. Bo C, Li R, Zhao JH, Feng S, Ji X, Xue F. Agent T-STROBE: A front-shifted STROBE paradigm for observational research.