# Mark McCarthy

#### List of Publications by Citations

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161 125,249 559 349 g-index h-index citations papers 612 149,613 14.3 7.45 avg, IF L-index ext. citations ext. papers

#	Paper	IF	Citations
559	Analysis of protein-coding genetic variation in 60,706 humans. <i>Nature</i> , <b>2016</b> , 536, 285-91	50.4	6940
558	Finding the missing heritability of complex diseases. <i>Nature</i> , <b>2009</b> , 461, 747-53	50.4	6084
557	The Genotype-Tissue Expression (GTEx) project. <i>Nature Genetics</i> , <b>2013</b> , 45, 580-5	36.3	4179
556	A common variant in the FTO gene is associated with body mass index and predisposes to childhood and adult obesity. <i>Science</i> , <b>2007</b> , 316, 889-94	33.3	3294
555	Human genomics. The Genotype-Tissue Expression (GTEx) pilot analysis: multitissue gene regulation in humans. <i>Science</i> , <b>2015</b> , 348, 648-60	33.3	3242
554	Biological, clinical and population relevance of 95 loci for blood lipids. <i>Nature</i> , <b>2010</b> , 466, 707-13	50.4	2742
553	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , <b>2015</b> , 518, 197-206	50.4	2687
552	Age-related clonal hematopoiesis associated with adverse outcomes. <i>New England Journal of Medicine</i> , <b>2014</b> , 371, 2488-98	59.2	2314
551	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. <i>Nature Genetics</i> , <b>2010</b> , 42, 937-48	36.3	2267
550	Genome-wide association studies for complex traits: consensus, uncertainty and challenges. <i>Nature Reviews Genetics</i> , <b>2008</b> , 9, 356-69	30.1	2126
549	Discovery and refinement of loci associated with lipid levels. <i>Nature Genetics</i> , <b>2013</b> , 45, 1274-1283	36.3	1904
548	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes. <i>Science</i> , <b>2007</b> , 316, 1336-41	33.3	1823
547	10 Years of GWAS Discovery: Biology, Function, and Translation. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 5-22	11	1651
546	Five years of GWAS discovery. American Journal of Human Genetics, 2012, 90, 7-24	11	1635
545	Hundreds of variants clustered in genomic loci and biological pathways affect human height. <i>Nature</i> , <b>2010</b> , 467, 832-8	50.4	1514
544	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. <i>Nature Genetics</i> , <b>2008</b> , 40, 638-45	36.3	1496
543	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. <i>Nature Genetics</i> , <b>2012</b> , 44, 981-90	36.3	1482

# (2009-2010)

542	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. <i>Nature Genetics</i> , <b>2010</b> , 42, 579-89	36.3	1449
541	A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , <b>2016</b> , 48, 1279-83	36.3	1447
540	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. <i>Nature Genetics</i> , <b>2009</b> , 41, 25-34	36.3	1368
539	Defining the role of common variation in the genomic and biological architecture of adult human height. <i>Nature Genetics</i> , <b>2014</b> , 46, 1173-86	36.3	1339
538	Transcriptome and genome sequencing uncovers functional variation in humans. <i>Nature</i> , <b>2013</b> , 501, 50	6- <b>5</b> 1.4	1323
537	Common genetic determinants of vitamin D insufficiency: a genome-wide association study. <i>Lancet, The,</i> <b>2010</b> , 376, 180-8	40	1183
536	Large-scale association analysis identifies new risk loci for coronary artery disease. <i>Nature Genetics</i> , <b>2013</b> , 45, 25-33	36.3	1172
535	Genome-wide association analysis identifies 13 new risk loci for schizophrenia. <i>Nature Genetics</i> , <b>2013</b> , 45, 1150-9	36.3	1153
534	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. <i>Nature Genetics</i> , <b>2007</b> , 39, 1329-37	36.3	1130
533	Common variants near MC4R are associated with fat mass, weight and risk of obesity. <i>Nature Genetics</i> , <b>2008</b> , 40, 768-75	36.3	1048
532	Genome-wide association study identifies eight loci associated with blood pressure. <i>Nature Genetics</i> , <b>2009</b> , 41, 666-76	36.3	970
531	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , <b>2015</b> , 518, 187-196	50.4	920
530	Metabolic profiling reveals a contribution of gut microbiota to fatty liver phenotype in insulin-resistant mice. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2006</b> , 103, 12511-6	11.5	854
529	A large-scale genome-wide association study of Asian populations uncovers genetic factors influencing eight quantitative traits. <i>Nature Genetics</i> , <b>2009</b> , 41, 527-34	36.3	822
528	Conditional and joint multiple-SNP analysis of GWAS summary statistics identifies additional variants influencing complex traits. <i>Nature Genetics</i> , <b>2012</b> , 44, 369-75, S1-3	36.3	813
527	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. <i>Nature Genetics</i> , <b>2014</b> , 46, 234-44	36.3	784
526	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. <i>Nature Genetics</i> , <b>2010</b> , 42, 949-60	36.3	724
525	Loci influencing lipid levels and coronary heart disease risk in 16 European population cohorts.  Nature Genetics, <b>2009</b> , 41, 47-55	36.3	708

524	The genetic architecture of type 2 diabetes. <i>Nature</i> , <b>2016</b> , 536, 41-47	50.4	704
523	Genomics, type 2 diabetes, and obesity. New England Journal of Medicine, 2010, 363, 2339-50	59.2	696
522	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. <i>Nature Genetics</i> , <b>2018</b> , 50, 1505-1513	36.3	675
521	Genome-wide association analysis identifies 20 loci that influence adult height. <i>Nature Genetics</i> , <b>2008</b> , 40, 575-83	36.3	654
520	Interaction between ERAP1 and HLA-B27 in ankylosing spondylitis implicates peptide handling in the mechanism for HLA-B27 in disease susceptibility. <i>Nature Genetics</i> , <b>2011</b> , 43, 761-7	36.3	646
519	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. <i>Nature</i> , <b>2010</b> , 464, 713-20	50.4	639
518	Identification of seven loci affecting mean telomere length and their association with disease. <i>Nature Genetics</i> , <b>2013</b> , 45, 422-7, 427e1-2	36.3	624
5 <del>1</del> 7	Large-scale association analyses identify new loci influencing glycemic traits and provide insight into the underlying biological pathways. <i>Nature Genetics</i> , <b>2012</b> , 44, 991-1005	36.3	621
516	A genome-wide approach accounting for body mass index identifies genetic variants influencing fasting glycemic traits and insulin resistance. <i>Nature Genetics</i> , <b>2012</b> , 44, 659-69	36.3	615
515	Large-scale association studies of variants in genes encoding the pancreatic beta-cell KATP channel subunits Kir6.2 (KCNJ11) and SUR1 (ABCC8) confirm that the KCNJ11 E23K variant is associated with type 2 diabetes. <i>Diabetes</i> , <b>2003</b> , 52, 568-72	0.9	614
5 <sup>1</sup> 4	Common variants associated with plasma triglycerides and risk for coronary artery disease. <i>Nature Genetics</i> , <b>2013</b> , 45, 1345-52	36.3	597
513	Causal relationship between obesity and vitamin D status: bi-directional Mendelian randomization analysis of multiple cohorts. <i>PLoS Medicine</i> , <b>2013</b> , 10, e1001383	11.6	592
512	Genome-wide association analysis of metabolic traits in a birth cohort from a founder population. <i>Nature Genetics</i> , <b>2009</b> , 41, 35-46	36.3	588
511	Variants in MTNR1B influence fasting glucose levels. <i>Nature Genetics</i> , <b>2009</b> , 41, 77-81	36.3	584
510	Sequence variants at CHRNB3-CHRNA6 and CYP2A6 affect smoking behavior. <i>Nature Genetics</i> , <b>2010</b> , 42, 448-53	36.3	582
509	Mapping cis- and trans-regulatory effects across multiple tissues in twins. <i>Nature Genetics</i> , <b>2012</b> , 44, 10	)8 <del>4</del> 693	572
508	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. <i>Nature Genetics</i> , <b>2010</b> , 42, 142-8	36.3	527
507	Genome-wide association study for early-onset and morbid adult obesity identifies three new risk loci in European populations. <i>Nature Genetics</i> , <b>2009</b> , 41, 157-9	36.3	521

506	Epigenome-wide association study of body mass index, and the adverse outcomes of adiposity. <i>Nature</i> , <b>2017</b> , 541, 81-86	50.4	511
505	Epigenome-wide scans identify differentially methylated regions for age and age-related phenotypes in a healthy ageing population. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002629	6	501
504	Meta-analysis of 28,141 individuals identifies common variants within five new loci that influence uric acid concentrations. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000504	6	495
503	Meta-analysis of genome-wide association studies identifies eight new loci for type 2 diabetes in east Asians. <i>Nature Genetics</i> , <b>2011</b> , 44, 67-72	36.3	475
502	Genetic Loci associated with C-reactive protein levels and risk of coronary heart disease. <i>JAMA - Journal of the American Medical Association</i> , <b>2009</b> , 302, 37-48	27.4	459
501	Assessment of cumulative evidence on genetic associations: interim guidelines. <i>International Journal of Epidemiology</i> , <b>2008</b> , 37, 120-32	7.8	451
500	Genome-wide association study identifies multiple loci influencing human serum metabolite levels. <i>Nature Genetics</i> , <b>2012</b> , 44, 269-76	36.3	441
499	Genome-wide meta-analysis identifies 11 new loci for anthropometric traits and provides insights into genetic architecture. <i>Nature Genetics</i> , <b>2013</b> , 45, 501-12	36.3	437
498	Genome-wide association study identifies loci influencing concentrations of liver enzymes in plasma. <i>Nature Genetics</i> , <b>2011</b> , 43, 1131-8	36.3	415
497	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. <i>Diabetes</i> , <b>2017</b> , 66, 288	38-2902	2 414
496	Rare and low-frequency coding variants alter human adult height. <i>Nature</i> , <b>2017</b> , 542, 186-190	50.4	412
495	Genome-wide association study of ulcerative colitis identifies three new susceptibility loci, including the HNF4A region. <i>Nature Genetics</i> , <b>2009</b> , 41, 1330-4	36.3	411
494	What makes a good genetic association study?. Lancet, The, 2005, 366, 1315-23	40	408
493	Genome-wide association study in individuals of South Asian ancestry identifies six new type 2 diabetes susceptibility loci. <i>Nature Genetics</i> , <b>2011</b> , 43, 984-9	36.3	406
492	Parent-of-origin-specific allelic associations among 106 genomic loci for age at menarche. <i>Nature</i> , <b>2014</b> , 514, 92-97	50.4	401
491	The metabochip, a custom genotyping array for genetic studies of metabolic, cardiovascular, and anthropometric traits. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002793	6	395
490	Genome-wide association scan meta-analysis identifies three Loci influencing adiposity and fat distribution. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000508	6	393
489	Causal relationships among the gut microbiome, short-chain fatty acids and metabolic diseases. <i>Nature Genetics</i> , <b>2019</b> , 51, 600-605	36.3	378

488	Pancreatic islet enhancer clusters enriched in type 2 diabetes risk-associated variants. <i>Nature Genetics</i> , <b>2014</b> , 46, 136-143	36.3	366
487	Loss-of-function mutations in SLC30A8 protect against type 2 diabetes. <i>Nature Genetics</i> , <b>2014</b> , 46, 357-6	5 <b>3</b> 6.3	351
486	Bayesian refinement of association signals for 14 loci in 3 common diseases. <i>Nature Genetics</i> , <b>2012</b> , 44, 1294-301	36.3	347
485	Human Lell transcriptome analysis uncovers lncRNAs that are tissue-specific, dynamically regulated, and abnormally expressed in type 2 diabetes. <i>Cell Metabolism</i> , <b>2012</b> , 16, 435-48	24.6	345
484	The architecture of gene regulatory variation across multiple human tissues: the MuTHER study. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1002003	6	336
483	Genomic inflation factors under polygenic inheritance. <i>European Journal of Human Genetics</i> , <b>2011</b> , 19, 807-12	5.3	335
482	A genome-wide association study identifies protein quantitative trait loci (pQTLs). <i>PLoS Genetics</i> , <b>2008</b> , 4, e1000072	6	331
481	A common variant of HMGA2 is associated with adult and childhood height in the general population. <i>Nature Genetics</i> , <b>2007</b> , 39, 1245-50	36.3	330
480	Coding Variation in ANGPTL4, LPL, and SVEP1 and the Risk of Coronary Disease. <i>New England Journal of Medicine</i> , <b>2016</b> , 374, 1134-44	59.2	325
479	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. <i>Nature</i> , <b>2011</b> , 478, 97-102	50.4	322
478	Concept, design and implementation of a cardiovascular gene-centric 50 k SNP array for large-scale genomic association studies. <i>PLoS ONE</i> , <b>2008</b> , 3, e3583	3.7	321
477	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. <i>Nature Genetics</i> , <b>2011</b> , 43, 117-20	36.3	319
476	Common variants at 10 genomic loci influence hemoglobin A(C) levels via glycemic and nonglycemic pathways. <i>Diabetes</i> , <b>2010</b> , 59, 3229-39	0.9	314
475	The human pancreatic islet transcriptome: expression of candidate genes for type 1 diabetes and the impact of pro-inflammatory cytokines. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002552	6	313
474	Integrative genomic analysis implicates limited peripheral adipose storage capacity in the pathogenesis of human insulin resistance. <i>Nature Genetics</i> , <b>2017</b> , 49, 17-26	36.3	312
473	Exome-wide association study of plasma lipids in >300,000 individuals. <i>Nature Genetics</i> , <b>2017</b> , 49, 1758-	1 <b>36</b> .6	310
472	FTO genotype is associated with phenotypic variability of body mass index. <i>Nature</i> , <b>2012</b> , 490, 267-72	50.4	304
471	Common variants in WFS1 confer risk of type 2 diabetes. <i>Nature Genetics</i> , <b>2007</b> , 39, 951-3	36.3	296

# (2016-2013)

470	Improved detection of common variants associated with schizophrenia by leveraging pleiotropy with cardiovascular-disease risk factors. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 197-209	11	293
469	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. <i>Nature Genetics</i> , <b>2015</b> , 47, 1415-25	36.3	292
468	A genome-wide association meta-analysis identifies new childhood obesity loci. <i>Nature Genetics</i> , <b>2012</b> , 44, 526-31	36.3	292
467	Mapping of a gene for type 2 diabetes associated with an insulin secretion defect by a genome scan in Finnish families. <i>Nature Genetics</i> , <b>1996</b> , 14, 90-4	36.3	<b>2</b> 90
466	Genome-wide association identifies nine common variants associated with fasting proinsulin levels and provides new insights into the pathophysiology of type 2 diabetes. <i>Diabetes</i> , <b>2011</b> , 60, 2624-34	0.9	285
465	Rare MTNR1B variants impairing melatonin receptor 1B function contribute to type 2 diabetes. <i>Nature Genetics</i> , <b>2012</b> , 44, 297-301	36.3	279
464	Epigenome-wide association of DNA methylation markers in peripheral blood from Indian Asians and Europeans with incident type 2 diabetes: a nested case-control study. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2015</b> , 3, 526-534	18.1	277
463	Sex-stratified genome-wide association studies including 270,000 individuals show sexual dimorphism in genetic loci for anthropometric traits. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003500	6	277
462	A genomewide scan for loci predisposing to type 2 diabetes in a U.K. population (the Diabetes UK Warren 2 Repository): analysis of 573 pedigrees provides independent replication of a susceptibility locus on chromosome 1q. <i>American Journal of Human Genetics</i> , <b>2001</b> , 69, 553-69	11	271
461	Global analysis of DNA methylation variation in adipose tissue from twins reveals links to disease-associated variants in distal regulatory elements. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 876-90	11	269
460	Genome-wide associations for birth weight and correlations with adult disease. <i>Nature</i> , <b>2016</b> , 538, 248	-25⁄2.4	266
459	Genome-wide association studies: potential next steps on a genetic journey. <i>Human Molecular Genetics</i> , <b>2008</b> , 17, R156-65	5.6	263
458	Common variation in the FTO gene alters diabetes-related metabolic traits to the extent expected given its effect on BMI. <i>Diabetes</i> , <b>2008</b> , 57, 1419-26	0.9	260
457	Genomic analyses identify hundreds of variants associated with age at menarche and support a role for puberty timing in cancer risk. <i>Nature Genetics</i> , <b>2017</b> , 49, 834-841	36.3	257
456	Research capacity. Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-8	33.3	256
455	Linkage and association of insulin gene VNTR regulatory polymorphism with polycystic ovary syndrome. <i>Lancet, The</i> , <b>1997</b> , 349, 986-90	40	254
454	Identification of an imprinted master trans regulator at the KLF14 locus related to multiple metabolic phenotypes. <i>Nature Genetics</i> , <b>2011</b> , 43, 561-4	36.3	253
453	The genetics of blood pressure regulation and its target organs from association studies in 342,415 individuals. <i>Nature Genetics</i> , <b>2016</b> , 48, 1171-1184	36.3	251

452	Variation in TCF7L2 influences therapeutic response to sulfonylureas: a GoDARTs study. <i>Diabetes</i> , <b>2007</b> , 56, 2178-82	0.9	251
45 <sup>1</sup>	Association of vitamin D status with arterial blood pressure and hypertension risk: a mendelian randomisation study. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2014</b> , 2, 719-29	18.1	250
450	Two new Loci for body-weight regulation identified in a joint analysis of genome-wide association studies for early-onset extreme obesity in French and german study groups. <i>PLoS Genetics</i> , <b>2010</b> , 6, e10	00916	250
449	Novel insights into the genetics of smoking behaviour, lung function, and chronic obstructive pulmonary disease (UK BiLEVE): a genetic association study in UK Biobank. <i>Lancet Respiratory Medicine,the</i> , <b>2015</b> , 3, 769-81	35.1	245
448	Assessing the combined impact of 18 common genetic variants of modest effect sizes on type 2 diabetes risk. <i>Diabetes</i> , <b>2008</b> , 57, 3129-35	0.9	245
447	Distribution and medical impact of loss-of-function variants in the Finnish founder population. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004494	6	243
446	Using an atlas of gene regulation across 44 human tissues to inform complex disease- and trait-associated variation. <i>Nature Genetics</i> , <b>2018</b> , 50, 956-967	36.3	239
445	Regulation of Fto/Ftm gene expression in mice and humans. <i>American Journal of Physiology - Regulatory Integrative and Comparative Physiology</i> , <b>2008</b> , 294, R1185-96	3.2	238
444	Impact of type 2 diabetes susceptibility variants on quantitative glycemic traits reveals mechanistic heterogeneity. <i>Diabetes</i> , <b>2014</b> , 63, 2158-71	0.9	235
443	New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. <i>Nature Genetics</i> , <b>2013</b> , 45, 76-82	36.3	232
442	The impact of low-frequency and rare variants on lipid levels. <i>Nature Genetics</i> , <b>2015</b> , 47, 589-97	36.3	229
441	Association Between Low-Density Lipoprotein Cholesterol-Lowering Genetic Variants and Risk of Type 2 Diabetes: A Meta-analysis. <i>JAMA - Journal of the American Medical Association</i> , <b>2016</b> , 316, 1383-1	39 <sup>7</sup> 1 <sup>4</sup>	225
440	Detection of human adaptation during the past 2000 years. Science, 2016, 354, 760-764	33.3	224
439	Trans-ancestry genome-wide association study identifies 12 genetic loci influencing blood pressure and implicates a role for DNA methylation. <i>Nature Genetics</i> , <b>2015</b> , 47, 1282-1293	36.3	223
438	Impact of common genetic determinants of Hemoglobin A1c on type 2 diabetes risk and diagnosis in ancestrally diverse populations: A transethnic genome-wide meta-analysis. <i>PLoS Medicine</i> , <b>2017</b> , 14, e1002383	11.6	223
437	Association analysis of 6,736 U.K. subjects provides replication and confirms TCF7L2 as a type 2 diabetes susceptibility gene with a substantial effect on individual risk. <i>Diabetes</i> , <b>2006</b> , 55, 2640-4	0.9	222
436	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. <i>Nature Genetics</i> , <b>2018</b> , 50, 559-571	36.3	221
435	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005378	6	220

# (2010-2011)

434	gene (AUTS2) in the regulation of alcohol consumption. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2011</b> , 108, 7119-24	11.5	218
433	Genome-wide association of polycystic ovary syndrome implicates alterations in gonadotropin secretion in European ancestry populations. <i>Nature Communications</i> , <b>2015</b> , 6, 7502	17.4	214
432	Large-scale gene-centric meta-analysis across 39 studies identifies type 2 diabetes loci. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 410-25	11	214
431	Combining information from common type 2 diabetes risk polymorphisms improves disease prediction. <i>PLoS Medicine</i> , <b>2006</b> , 3, e374	11.6	214
430	Genetic Predisposition to an Impaired Metabolism of the Branched-Chain Amino Acids and Risk of Type 2 Diabetes: A Mendelian Randomisation Analysis. <i>PLoS Medicine</i> , <b>2016</b> , 13, e1002179	11.6	214
429	Detailed physiologic characterization reveals diverse mechanisms for novel genetic Loci regulating glucose and insulin metabolism in humans. <i>Diabetes</i> , <b>2010</b> , 59, 1266-75	0.9	211
428	Genetic loci influencing kidney function and chronic kidney disease. <i>Nature Genetics</i> , <b>2010</b> , 42, 373-5	36.3	205
427	Type 2 diabetes: new genes, new understanding. <i>Trends in Genetics</i> , <b>2008</b> , 24, 613-21	8.5	204
426	Common variants of the novel type 2 diabetes genes CDKAL1 and HHEX/IDE are associated with decreased pancreatic beta-cell function. <i>Diabetes</i> , <b>2007</b> , 56, 3101-4	0.9	203
425	Early life factors and blood pressure at age 31 years in the 1966 northern Finland birth cohort. <i>Hypertension</i> , <b>2004</b> , 44, 838-46	8.5	203
424	Genome-wide association analysis identifies three new susceptibility loci for childhood body mass index. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 389-403	5.6	202
423	A bivariate genome-wide approach to metabolic syndrome: STAMPEED consortium. <i>Diabetes</i> , <b>2011</b> , 60, 1329-39	0.9	194
422	Integrated genetic and epigenetic analysis identifies haplotype-specific methylation in the FTO type 2 diabetes and obesity susceptibility locus. <i>PLoS ONE</i> , <b>2010</b> , 5, e14040	3.7	193
421	Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer. <i>Nature</i> , <b>2013</b> , 493, 406-10	50.4	191
420	A coherent approach for analysis of the Illumina HumanMethylation450 BeadChip improves data quality and performance in epigenome-wide association studies. <i>Genome Biology</i> , <b>2015</b> , 16, 37	18.3	186
419	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. <i>Nature Genetics</i> , <b>2018</b> , 50, 26-41	36.3	186
418	Gene expression changes with age in skin, adipose tissue, blood and brain. <i>Genome Biology</i> , <b>2013</b> , 14, R75	18.3	185
417	Variants in ADCY5 and near CCNL1 are associated with fetal growth and birth weight. <i>Nature Genetics</i> , <b>2010</b> , 42, 430-5	36.3	184

416	Development of polycystic ovary syndrome: involvement of genetic and environmental factors. Journal of Developmental and Physical Disabilities, <b>2006</b> , 29, 278-85; discussion 286-90		184
415	Evidence that a locus for familial psoriasis maps to chromosome 4q. <i>Nature Genetics</i> , <b>1996</b> , 14, 231-3	36.3	182
414	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. <i>Nature Genetics</i> , <b>2019</b> , 51, 804-814	36.3	181
413	Trans-ancestry meta-analyses identify rare and common variants associated with blood pressure and hypertension. <i>Nature Genetics</i> , <b>2016</b> , 48, 1151-1161	36.3	181
412	A brief history of human disease genetics. <i>Nature</i> , <b>2020</b> , 577, 179-189	50.4	181
411	New loci for body fat percentage reveal link between adiposity and cardiometabolic disease risk. <i>Nature Communications</i> , <b>2016</b> , 7, 10495	17.4	180
410	Remapping the insulin gene/IDDM2 locus in type 1 diabetes. <i>Diabetes</i> , <b>2004</b> , 53, 1884-9	0.9	180
409	Genome-wide association study identifies variants in TMPRSS6 associated with hemoglobin levels. <i>Nature Genetics</i> , <b>2009</b> , 41, 1170-2	36.3	179
408	Dissection of the genetics of ParkinsonN disease identifies an additional association 5Nof SNCA and multiple associated haplotypes at 17q21. <i>Human Molecular Genetics</i> , <b>2011</b> , 20, 345-53	5.6	178
407	Genome-wide association study in 79,366 European-ancestry individuals informs the genetic architecture of 25-hydroxyvitamin D levels. <i>Nature Communications</i> , <b>2018</b> , 9, 260	17.4	174
406	RNA sequencing identifies dysregulation of the human pancreatic islet transcriptome by the saturated fatty acid palmitate. <i>Diabetes</i> , <b>2014</b> , 63, 1978-93	0.9	174
405	Reduced insulin exocytosis in human pancreatic Etells with gene variants linked to type 2 diabetes. <i>Diabetes</i> , <b>2012</b> , 61, 1726-33	0.9	174
404	Genome-wide association studies in type 2 diabetes. Current Diabetes Reports, 2009, 9, 164-71	5.6	174
403	Human genomics. Effect of predicted protein-truncating genetic variants on the human transcriptome. <i>Science</i> , <b>2015</b> , 348, 666-9	33.3	170
402	Whole-exome sequencing identifies rare and low-frequency coding variants associated with LDL cholesterol. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 233-45	11	170
401	Large-scale genome-wide meta-analysis of polycystic ovary syndrome suggests shared genetic architecture for different diagnosis criteria. <i>PLoS Genetics</i> , <b>2018</b> , 14, e1007813	6	166
400	The genetic and epigenetic basis of type 2 diabetes and obesity. <i>Clinical Pharmacology and Therapeutics</i> , <b>2012</b> , 92, 707-15	6.1	162
399	Stratifying type 2 diabetes cases by BMI identifies genetic risk variants in LAMA1 and enrichment for risk variants in lean compared to obese cases. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002741	6	162

398	Pharmacogenetic meta-analysis of genome-wide association studies of LDL cholesterol response to statins. <i>Nature Communications</i> , <b>2014</b> , 5, 5068	17.4	160
397	Evaluation of common variants in the six known maturity-onset diabetes of the young (MODY) genes for association with type 2 diabetes. <i>Diabetes</i> , <b>2007</b> , 56, 685-93	0.9	160
396	Meta-analysis and a large association study confirm a role for calpain-10 variation in type 2 diabetes susceptibility. <i>American Journal of Human Genetics</i> , <b>2003</b> , 73, 1208-12	11	155
395	Studies of association between the gene for calpain-10 and type 2 diabetes mellitus in the United Kingdom. <i>American Journal of Human Genetics</i> , <b>2001</b> , 69, 544-52	11	154
394	Genetic evidence that raised sex hormone binding globulin (SHBG) levels reduce the risk of type 2 diabetes. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 535-44	5.6	150
393	Genetic Evidence for Causal Relationships Between Maternal Obesity-Related Traits and Birth Weight. <i>JAMA - Journal of the American Medical Association</i> , <b>2016</b> , 315, 1129-40	27.4	149
392	Gene-lifestyle interaction and type 2 diabetes: the EPIC interact case-cohort study. <i>PLoS Medicine</i> , <b>2014</b> , 11, e1001647	11.6	149
391	Systematic Evaluation of Pleiotropy Identifies 6 Further Loci Associated With Coronary Artery Disease. <i>Journal of the American College of Cardiology</i> , <b>2017</b> , 69, 823-836	15.1	146
390	The miRNA profile of human pancreatic islets and beta-cells and relationship to type 2 diabetes pathogenesis. <i>PLoS ONE</i> , <b>2013</b> , 8, e55272	3.7	146
389	Diabetes and Cause-Specific Mortality in Mexico City. New England Journal of Medicine, 2016, 375, 1961	- <del>1,9</del> 721	144
388	The role of adiposity in cardiometabolic traits: a Mendelian randomization analysis. <i>PLoS Medicine</i> , <b>2013</b> , 10, e1001474	11.6	144
387	Association between common variation at the FTO locus and changes in body mass index from infancy to late childhood: the complex nature of genetic association through growth and development. <i>PLoS Genetics</i> , <b>2011</b> , 7, e1001307	6	141
386	Exploring the developmental overnutrition hypothesis using parental-offspring associations and FTO as an instrumental variable. <i>PLoS Medicine</i> , <b>2008</b> , 5, e33	11.6	139
385	Genome-wide association and longitudinal analyses reveal genetic loci linking pubertal height growth, pubertal timing and childhood adiposity. <i>Human Molecular Genetics</i> , <b>2013</b> , 22, 2735-47	5.6	138
384	Transcript Expression Data from Human Islets Links Regulatory Signals from Genome-Wide Association Studies for Type 2 Diabetes and Glycemic Traits to Their Downstream Effectors. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005694	6	138
383	A genome-wide association search for type 2 diabetes genes in African Americans. <i>PLoS ONE</i> , <b>2012</b> ,	3.7	138
	7, e29202	<i>J</i> /	
382	Exposing the exposures responsible for type 2 diabetes and obesity. <i>Science</i> , <b>2016</b> , 354, 69-73	33.3	138

380	Reduced-function SLC22A1 polymorphisms encoding organic cation transporter 1 and glycemic response to metformin: a GoDARTS study. <i>Diabetes</i> , <b>2009</b> , 58, 1434-9	0.9	132
379	eVOC: a controlled vocabulary for unifying gene expression data. <i>Genome Research</i> , <b>2003</b> , 13, 1222-30	9.7	132
378	Exome sequencing of 20,791 cases of type 2 diabetes and 24,440 controls. <i>Nature</i> , <b>2019</b> , 570, 71-76	50.4	129
377	Genome-wide association study identifies a novel locus contributing to type 2 diabetes susceptibility in Sikhs of Punjabi origin from India. <i>Diabetes</i> , <b>2013</b> , 62, 1746-55	0.9	129
376	Global adiposity rather than abnormal regional fat distribution characterizes women with polycystic ovary syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2008</b> , 93, 999-1004	5.6	129
375	Common genetic variants highlight the role of insulin resistance and body fat distribution in type 2 diabetes, independent of obesity. <i>Diabetes</i> , <b>2014</b> , 63, 4378-4387	0.9	127
374	Genome-wide association study identifies novel genetic variants contributing to variation in blood metabolite levels. <i>Nature Communications</i> , <b>2015</b> , 6, 7208	17.4	126
373	A central role for GRB10 in regulation of islet function in man. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004235	6	124
372	Systematic assessment of etiology in adults with a clinical diagnosis of young-onset type 2 diabetes is a successful strategy for identifying maturity-onset diabetes of the young. <i>Diabetes Care</i> , <b>2012</b> , 35, 1206-12	14.6	122
371	Using human genetics to understand the disease impacts of testosterone in men and women. <i>Nature Medicine</i> , <b>2020</b> , 26, 252-258	50.5	121
370	Manifestations of metabolic syndrome after hypertensive pregnancy. <i>Hypertension</i> , <b>2004</b> , 43, 825-31	8.5	121
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368	Human metabolic profiles are stably controlled by genetic and environmental variation. <i>Molecular Systems Biology</i> , <b>2011</b> , 7, 525	12.2	119
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366	A genome-wide association meta-analysis of circulating sex hormone-binding globulin reveals multiple Loci implicated in sex steroid hormone regulation. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002805	6	116
365	Genetic determinants of height growth assessed longitudinally from infancy to adulthood in the northern Finland birth cohort 1966. <i>PLoS Genetics</i> , <b>2009</b> , 5, e1000409	6	113
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361	Distinct developmental profile of lower-body adipose tissue defines resistance against obesity-associated metabolic complications. <i>Diabetes</i> , <b>2014</b> , 63, 3785-97	0.9	107
360	Genome-wide meta-analysis of 241,258 adults accounting for smoking behaviour identifies novel loci for obesity traits. <i>Nature Communications</i> , <b>2017</b> , 8, 14977	17.4	105
359	Cytotoxic T lymphocyte antigen-4 (CTLA-4) gene polymorphism confers susceptibility to thyroid associated orbitopathy. <i>Lancet, The</i> , <b>1999</b> , 354, 743-4	40	105
358	Common variants at 6q22 and 17q21 are associated with intracranial volume. <i>Nature Genetics</i> , <b>2012</b> , 44, 539-44	36.3	104
357	Genome-wide physical activity interactions in adiposity - A meta-analysis of 200,452 adults. <i>PLoS Genetics</i> , <b>2017</b> , 13, e1006528	6	103
356	Painting a new picture of personalised medicine for diabetes. <i>Diabetologia</i> , <b>2017</b> , 60, 793-799	10.3	102
355	Type 2 diabetes TCF7L2 risk genotypes alter birth weight: a study of 24,053 individuals. <i>American Journal of Human Genetics</i> , <b>2007</b> , 80, 1150-61	11	100
354	Candidate genes in polycystic ovary syndrome. Human Reproduction Update, 2001, 7, 405-10	15.8	100
353	The genetics of diabetic complications. <i>Nature Reviews Nephrology</i> , <b>2015</b> , 11, 277-87	14.9	99
353 352	The genetics of diabetic complications. <i>Nature Reviews Nephrology</i> , <b>2015</b> , 11, 277-87  Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67	14.9 5.6	99 99
	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes</i>	. ,	
352	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67  The power of gene-based rare variant methods to detect disease-associated variation and test	5.6	99
352 351	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67  The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005165  Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. <i>Human Molecular</i>	5.6	99
35 <sup>2</sup> 35 <sup>1</sup> 35 <sup>0</sup>	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67  The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005165  Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 742-756  Adiposity-related heterogeneity in patterns of type 2 diabetes susceptibility observed in	5.6 6 5.6	99 98 98
35 <sup>2</sup> 35 <sup>1</sup> 35 <sup>0</sup>	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67  The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005165  Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 742-756  Adiposity-related heterogeneity in patterns of type 2 diabetes susceptibility observed in genome-wide association data. <i>Diabetes</i> , <b>2009</b> , 58, 505-10	5.6 6 5.6 0.9	99 98 98 98
35 <sup>2</sup> 35 <sup>1</sup> 35 <sup>0</sup> 349	Growing evidence for diabetes susceptibility genes from genome scan data. <i>Current Diabetes Reports</i> , <b>2003</b> , 3, 159-67  The power of gene-based rare variant methods to detect disease-associated variation and test hypotheses about complex disease. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1005165  Genome-wide association study of offspring birth weight in 86 577 women identifies five novel loci and highlights maternal genetic effects that are independent of fetal genetics. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 742-756  Adiposity-related heterogeneity in patterns of type 2 diabetes susceptibility observed in genome-wide association data. <i>Diabetes</i> , <b>2009</b> , 58, 505-10  TCF7L2: the biggest story in diabetes genetics since HLA?. <i>Diabetologia</i> , <b>2007</b> , 50, 1-4  Adiposity as a cause of cardiovascular disease: a Mendelian randomization study. <i>International</i>	5.6 6 5.6 0.9	99 98 98 98 98

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343	Mendelian randomization studies do not support a causal role for reduced circulating adiponectin levels in insulin resistance and type 2 diabetes. <i>Diabetes</i> , <b>2013</b> , 62, 3589-98	0.9	95
342	Genetic Risk Scores for Diabetes Diagnosis and Precision Medicine. <i>Endocrine Reviews</i> , <b>2019</b> , 40, 1500-1	5 <b>2</b> 70.2	94
341	Common variants at 12q15 and 12q24 are associated with infant head circumference. <i>Nature Genetics</i> , <b>2012</b> , 44, 532-538	36.3	94
340	Structural forms of the human amylase locus and their relationships to SNPs, haplotypes and obesity. <i>Nature Genetics</i> , <b>2015</b> , 47, 921-5	36.3	92
339	Hnf1alpha (MODY3) controls tissue-specific transcriptional programs and exerts opposed effects on cell growth in pancreatic islets and liver. <i>Molecular and Cellular Biology</i> , <b>2009</b> , 29, 2945-59	4.8	92
338	miR-375 gene dosage in pancreatic Etells: implications for regulation of Etell mass and biomarker development. <i>Journal of Molecular Medicine</i> , <b>2015</b> , 93, 1159-69	5.5	90
337	Identification of type 2 diabetes loci in 433,540 East Asian individuals. <i>Nature</i> , <b>2020</b> , 582, 240-245	50.4	89
336	Comprehensive human adipose tissue mRNA and microRNA endogenous control selection for quantitative real-time-PCR normalization. <i>Obesity</i> , <b>2011</b> , 19, 888-92	8	89
335	Assessment of high-sensitivity C-reactive protein levels as diagnostic discriminator of maturity-onset diabetes of the young due to HNF1A mutations. <i>Diabetes Care</i> , <b>2010</b> , 33, 1919-24	14.6	89
334	An evaluation of HapMap sample size and tagging SNP performance in large-scale empirical and simulated data sets. <i>Nature Genetics</i> , <b>2005</b> , 37, 1320-2	36.3	88
333	Insights into the molecular mechanism for type 2 diabetes susceptibility at the KCNQ1 locus from temporal changes in imprinting status in human islets. <i>Diabetes</i> , <b>2013</b> , 62, 987-92	0.9	87
332	Interrogating type 2 diabetes genome-wide association data using a biological pathway-based approach. <i>Diabetes</i> , <b>2009</b> , 58, 1463-7	0.9	87
331	Tensor decomposition for multiple-tissue gene expression experiments. <i>Nature Genetics</i> , <b>2016</b> , 48, 109	431009	87
330	Increased 5 alpha-reductase activity and adrenocortical drive in women with polycystic ovary syndrome. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2009</b> , 94, 3558-66	5.6	86
329	An integrated epigenomic analysis for type 2 diabetes susceptibility loci in monozygotic twins. <i>Nature Communications</i> , <b>2014</b> , 5, 5719	17.4	85
328	Large-scale analysis of the relationship between CYP11A promoter variation, polycystic ovarian syndrome, and serum testosterone. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 2408-13	5.6	84
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324	Regulatory variants at KLF14 influence type 2 diabetes risk via a female-specific effect on adipocyte size and body composition. <i>Nature Genetics</i> , <b>2018</b> , 50, 572-580	36.3	82	
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322	Total zinc intake may modify the glucose-raising effect of a zinc transporter (SLC30A8) variant: a 14-cohort meta-analysis. <i>Diabetes</i> , <b>2011</b> , 60, 2407-16	0.9	81	
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320	Common genetic variation near melatonin receptor MTNR1B contributes to raised plasma glucose and increased risk of type 2 diabetes among Indian Asians and European Caucasians. <i>Diabetes</i> , <b>2009</b> , 58, 2703-8	0.9	80	
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318	Reply to Dlouha et al. European Journal of Human Genetics, 2010, 18, 1275-1275	5.3	78	
317	Metabolic cardiovascular disease risk factors in women with self-reported symptoms of oligomenorrhea and/or hirsutism: Northern Finland Birth Cohort 1966 Study. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 2114-8	5.6	78	
316	A novel common variant in DCST2 is associated with length in early life and height in adulthood. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1155-68	5.6	77	
315	A genomic approach to therapeutic target validation identifies a glucose-lowering GLP1R variant protective for coronary heart disease. <i>Science Translational Medicine</i> , <b>2016</b> , 8, 341ra76	17.5	77	
314	Identification and functional characterization of G6PC2 coding variants influencing glycemic traits define an effector transcript at the G6PC2-ABCB11 locus. <i>PLoS Genetics</i> , <b>2015</b> , 11, e1004876	6	76	
313	Mouse-human experimental epigenetic analysis unmasks dietary targets and genetic liability for diabetic phenotypes. <i>Cell Metabolism</i> , <b>2015</b> , 21, 138-49	24.6	76	
312	Heritability of variation in glycaemic response to metformin: a genome-wide complex trait analysis. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2014</b> , 2, 481-7	18.1	76	
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309	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 865-884	11	74	

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307	An association study of bipolar mood disorder (type I) with the 5-HTTLPR serotonin transporter polymorphism in a human population isolate from Colombia. <i>Neuroscience Letters</i> , <b>2000</b> , 292, 199-202	3.3	73
306	Prevalence of polycystic ovaries in women with androgenic alopecia. <i>European Journal of Endocrinology</i> , <b>2003</b> , 149, 439-42	6.5	72
305	A Genome-Wide Association Study of Diabetic Kidney Disease in Subjects With Type 2 Diabetes. <i>Diabetes</i> , <b>2018</b> , 67, 1414-1427	0.9	71
304	Genetic inactivation of ANGPTL4 improves glucose homeostasis and is associated with reduced risk of diabetes. <i>Nature Communications</i> , <b>2018</b> , 9, 2252	17.4	71
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302	Association of FTO variants with BMI and fat mass in the self-contained population of Sorbs in Germany. <i>European Journal of Human Genetics</i> , <b>2010</b> , 18, 104-10	5.3	71
301	Relationship between birthweight and blood lipid concentrations in later life: evidence from the existing literature. <i>International Journal of Epidemiology</i> , <b>2003</b> , 32, 862-76	7.8	71
300	Association of the CpG methylation pattern of the proximal insulin gene promoter with type 1 diabetes. <i>PLoS ONE</i> , <b>2012</b> , 7, e36278	3.7	71
299	Population whole-genome bisulfite sequencing across two tissues highlights the environment as the principal source of human methylome variation. <i>Genome Biology</i> , <b>2015</b> , 16, 290	18.3	70
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296	Improving reporting standards for polygenic scores in risk prediction studies. <i>Nature</i> , <b>2021</b> , 591, 211-21	<b>9</b> 50.4	70
295	Integration of human pancreatic islet genomic data refines regulatory mechanisms at Type 2 Diabetes susceptibility loci. <i>ELife</i> , <b>2018</b> , 7,	8.9	70
294	A Genome-Wide Association Study of IVGTT-Based Measures of First-Phase Insulin Secretion Refines the Underlying Physiology of Type 2 Diabetes Variants. <i>Diabetes</i> , <b>2017</b> , 66, 2296-2309	0.9	69
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291	Distinct variants at LIN28B influence growth in height from birth to adulthood. <i>American Journal of Human Genetics</i> , <b>2010</b> , 86, 773-82	11	68

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289	Genome-Wide Association Study of Diabetic Kidney Disease Highlights Biology Involved in Glomerular Basement Membrane Collagen. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2019</b> , 30, 2000-2016	12.7	66
288	Genome-wide association study of sexual maturation in males and females highlights a role for body mass and menarche loci in male puberty. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 4452-64	5.6	66
287	Characterization of functional methylomes by next-generation capture sequencing identifies novel disease-associated variants. <i>Nature Communications</i> , <b>2015</b> , 6, 7211	17.4	66
286	Paradoxical lower serum triglyceride levels and higher type 2 diabetes mellitus susceptibility in obese individuals with the PNPLA3 148M variant. <i>PLoS ONE</i> , <b>2012</b> , 7, e39362	3.7	66
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284	Variability of gene expression profiles in human blood and lymphoblastoid cell lines. <i>BMC Genomics</i> , <b>2010</b> , 11, 96	4.5	64
283	Association between the T-381C polymorphism of the brain natriuretic peptide gene and risk of type 2 diabetes in human populations. <i>Human Molecular Genetics</i> , <b>2007</b> , 16, 1343-50	5.6	64
282	Genetics of type 2 diabetes. Current Opinion in Genetics and Development, 2007, 17, 239-44	4.9	64
281	Meta-analysis investigating associations between healthy diet and fasting glucose and insulin levels and modification by loci associated with glucose homeostasis in data from 15 cohorts. <i>American Journal of Epidemiology</i> , <b>2013</b> , 177, 103-15	3.8	63
280	Genetic variations in the gene encoding TFAP2B are associated with type 2 diabetes mellitus. <i>Journal of Human Genetics</i> , <b>2005</b> , 50, 283-292	4.3	63
279	Type 2 diabetes and obesity: genomics and the clinic. <i>Human Genetics</i> , <b>2011</b> , 130, 41-58	6.3	62
278	Evidence of inbreeding depression on human height. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002655	6	62
277	Analysis of multiple data sets reveals no association between the insulin gene variable number tandem repeat element and polycystic ovary syndrome or related traits. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2005</b> , 90, 2988-93	5.6	62
276	Genetics meets proteomics: perspectives for large population-based studies. <i>Nature Reviews Genetics</i> , <b>2021</b> , 22, 19-37	30.1	62
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263	A Global Overview of Precision Medicine in Type 2 Diabetes. <i>Diabetes</i> , <b>2018</b> , 67, 1911-1922	0.9	52
262	The Genetic Basis of Metabolic Disease. <i>Cell</i> , <b>2019</b> , 177, 146-161	56.2	51
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259	Nonfasting glucose, ischemic heart disease, and myocardial infarction: a Mendelian randomization study. <i>Journal of the American College of Cardiology</i> , <b>2012</b> , 59, 2356-65	15.1	50
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155	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci		21
155 154	Exploiting the GTEx resources to decipher the mechanisms at GWAS loci  Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033	14.6	21
	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2	14.6	
154	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033  Association analysis of 29,956 individuals confirms that a low-frequency variant at CCND2 halves	·	20
154	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033  Association analysis of 29,956 individuals confirms that a low-frequency variant at CCND2 halves the risk of type 2 diabetes by enhancing insulin secretion. <i>Diabetes</i> , <b>2015</b> , 64, 2279-85  Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal	0.9	20
154 153 152	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033  Association analysis of 29,956 individuals confirms that a low-frequency variant at CCND2 halves the risk of type 2 diabetes by enhancing insulin secretion. <i>Diabetes</i> , <b>2015</b> , 64, 2279-85  Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory mechanisms. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2070-2081  Genetic Predisposition to Type 2 Diabetes and Risk of Subclinical Atherosclerosis and	o.9 5.6	20 20 20 20
154 153 152 151	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033  Association analysis of 29,956 individuals confirms that a low-frequency variant at CCND2 halves the risk of type 2 diabetes by enhancing insulin secretion. <i>Diabetes</i> , <b>2015</b> , 64, 2279-85  Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory mechanisms. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2070-2081  Genetic Predisposition to Type 2 Diabetes and Risk of Subclinical Atherosclerosis and Cardiovascular Diseases Among 160,000 Chinese Adults. <i>Diabetes</i> , <b>2019</b> , 68, 2155-2164	o.9 5.6 o.9	20 20 20 20
154 153 152 151 150	Variation in the Plasma Membrane Monoamine Transporter (PMAT) (Encoded by ) and Organic Cation Transporter 1 (OCT1) (Encoded by ) and Gastrointestinal Intolerance to Metformin in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2019</b> , 42, 1027-1033  Association analysis of 29,956 individuals confirms that a low-frequency variant at CCND2 halves the risk of type 2 diabetes by enhancing insulin secretion. <i>Diabetes</i> , <b>2015</b> , 64, 2279-85  Transancestral fine-mapping of four type 2 diabetes susceptibility loci highlights potential causal regulatory mechanisms. <i>Human Molecular Genetics</i> , <b>2016</b> , 25, 2070-2081  Genetic Predisposition to Type 2 Diabetes and Risk of Subclinical Atherosclerosis and Cardiovascular Diseases Among 160,000 Chinese Adults. <i>Diabetes</i> , <b>2019</b> , 68, 2155-2164  TCF7L2 and diabetes: a tale of two tissues, and of two species. <i>Cell Metabolism</i> , <b>2013</b> , 17, 157-9  Genome-wide association scans for Type 2 diabetes: new insights into biology and therapy. <i>Trends</i>	<ul><li>0.9</li><li>5.6</li><li>0.9</li><li>24.6</li></ul>	20 20 20 20 20

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144	Cohort Profile: East London Genes & Health (ELGH), a community-based population genomics and health study in British Bangladeshi and British Pakistani people. <i>International Journal of Epidemiology</i> , <b>2020</b> , 49, 20-21i	7.8	19
143	The Early Growth Genetics (EGG) and EArly Genetics and Lifecourse Epidemiology (EAGLE) consortia: design, results and future prospects. <i>European Journal of Epidemiology</i> , <b>2019</b> , 34, 279-300	12.1	18
142	Developing a network view of type 2 diabetes risk pathways through integration of genetic, genomic and functional data. <i>Genome Medicine</i> , <b>2019</b> , 11, 19	14.4	18
141	Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts. <i>PLoS Medicine</i> , <b>2020</b> , 17, e1003149	11.6	18
140	Insights into islet development and biology through characterization of a human iPSC-derived endocrine pancreas model. <i>Islets</i> , <b>2016</b> , 8, 83-95	2	18
139	No association between insulin gene variation and adult metabolic phenotypes in a large Finnish birth cohort. <i>Diabetologia</i> , <b>2005</b> , 48, 886-91	10.3	18
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137	Fine-mapping of an expanded set of type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps		18
136	Relationship between E23K (an established type II diabetes-susceptibility variant within KCNJ11), polycystic ovary syndrome and androgen levels. <i>European Journal of Human Genetics</i> , <b>2007</b> , 15, 679-84	5.3	17
135	PASSIMan open source software system for managing information in biomedical studies. <i>BMC Bioinformatics</i> , <b>2007</b> , 8, 52	3.6	17
		\\	
134	Bone mineral density and risk of type 2 diabetes and coronary heart disease: A Mendelian randomization study. <i>Wellcome Open Research</i> , <b>2017</b> , 2, 68	4.8	17
134		· ·	17 17
	randomization study. Wellcome Open Research, 2017, 2, 68  A novel rare CUBN variant and three additional genes identified in Europeans with and without	· ·	
133	randomization study. Wellcome Open Research, 2017, 2, 68  A novel rare CUBN variant and three additional genes identified in Europeans with and without diabetes: results from an exome-wide association study of albuminuria. Diabetologia, 2019, 62, 292-305  Characterising -regulatory variation in the transcriptome of histologically normal and	; 10.3	17
133	randomization study. Wellcome Open Research, 2017, 2, 68  A novel rare CUBN variant and three additional genes identified in Europeans with and without diabetes: results from an exome-wide association study of albuminuria. Diabetologia, 2019, 62, 292-305  Characterising -regulatory variation in the transcriptome of histologically normal and tumour-derived pancreatic tissues. Gut, 2018, 67, 521-533	19.2	17

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82	Will the real disease gene please stand up?. <i>BMC Genetics</i> , <b>2005</b> , 6 Suppl 1, S66  Whole-genome sequencing to understand the genetic architecture of common gene expression	2.6	8
82	Will the real disease gene please stand up?. <i>BMC Genetics</i> , <b>2005</b> , 6 Suppl 1, S66  Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1504-12  Childhood cognitive ability moderates later-life manifestation of type 2 diabetes genetic risk.	2.6	8
82 81 80	Will the real disease gene please stand up?. <i>BMC Genetics</i> , <b>2005</b> , 6 Suppl 1, S66  Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1504-12  Childhood cognitive ability moderates later-life manifestation of type 2 diabetes genetic risk. <i>Health Psychology</i> , <b>2015</b> , 34, 915-9  Examining the candidacy of ghrelin as a gene responsible for variation in adult stature in a United Kingdom population with type 2 diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2007</b> ,	<ul><li>2.6</li><li>5.6</li><li>5</li></ul>	8       7       7
82 81 80	Will the real disease gene please stand up?. <i>BMC Genetics</i> , <b>2005</b> , 6 Suppl 1, S66  Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1504-12  Childhood cognitive ability moderates later-life manifestation of type 2 diabetes genetic risk. <i>Health Psychology</i> , <b>2015</b> , 34, 915-9  Examining the candidacy of ghrelin as a gene responsible for variation in adult stature in a United Kingdom population with type 2 diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2007</b> , 92, 2201-4  Application of eVOC: controlled vocabularies for unifying gene expression data. <i>Comptes Rendus</i> -	2.6 5.6 5	8 7 7 7 7 7
82 81 80 79 78	Will the real disease gene please stand up?. <i>BMC Genetics</i> , <b>2005</b> , 6 Suppl 1, S66  Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 1504-12  Childhood cognitive ability moderates later-life manifestation of type 2 diabetes genetic risk. <i>Health Psychology</i> , <b>2015</b> , 34, 915-9  Examining the candidacy of ghrelin as a gene responsible for variation in adult stature in a United Kingdom population with type 2 diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2007</b> , 92, 2201-4  Application of eVOC: controlled vocabularies for unifying gene expression data. <i>Comptes Rendus - Biologies</i> , <b>2003</b> , 326, 1089-96	2.6 5.6 5 1.4	8 7 7 7

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Multi-ancestry genetic study of type 2 diabetes highlights the power of diverse populations for discovery and translation <i>Nature Genetics</i> , <b>2022</b> ,	36.3	7
A Multi-omic Integrative Scheme Characterizes Tissues of Action at Loci Associated with Type 2 Diabetes. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 1011-1028	11	6
Global Analysis of DNA Methylation Variation in Adipose Tissue from Twins Reveals Links to Disease-Associated Variants in Distal Regulatory Elements. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 1158	11	6
Evaluation of common type 2 diabetes risk variants in a South Asian population of Sri Lankan descent. <i>PLoS ONE</i> , <b>2014</b> , 9, e98608	3.7	6
Can surgeons assess CT suitability for endovascular repair (EVAR) in ruptured abdominal aortic aneurysm? Implications for a ruptured EVAR trial. <i>CardioVascular and Interventional Radiology</i> , <b>2008</b> , 31, 865-9	2.7	6
Processes Underlying Glycemic Deterioration in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , <b>2021</b> , 44, 511-518	14.6	6
Cohort Profile: East London Genes & Health (ELGH), a community based population genomics and health study of British-Bangladeshi and British-Pakistani people		6
Toppar: an interactive browser for viewing association study results. <i>Bioinformatics</i> , <b>2018</b> , 34, 1922-192	47.2	5
Metabolic and reproductive characteristics of first-degree relatives of women with self-reported oligo-amenorrhoea and hirsutism. <i>Gynecological Endocrinology</i> , <b>2011</b> , 27, 630-5	2.4	5
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Integrative network analysis highlights biological processes underlying GLP-1 stimulated insulin secretion: A DIRECT study. <i>PLoS ONE</i> , <b>2018</b> , 13, e0189886	3.7	5
Analysis of overlapping genetic association in type 1 and type 2 diabetes. <i>Diabetologia</i> , <b>2021</b> , 64, 1342-7	1 <b>3</b> 473	5
Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes. <i>Nature Communications</i> , <b>2021</b> , 12, 3505	17.4	5
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Identification of 371 genetic variants for age at first sex and birth linked to externalising behaviour. <i>Nature Human Behaviour</i> , <b>2021</b> ,	12.8	5
	discovery and translation <i>Nature Genetics</i> , 2022,  A Multi-omic Integrative Scheme Characterizes Tissues of Action at Loci Associated with Type 2 Diabetes. <i>American Journal of Human Genetics</i> , 2020, 107, 1011-1028  Global Analysis of DNA Methylation Variation in Adipose Tissue from Twins Reveals Links to Disease-Associated Variants in Distal Regulatory Elements. <i>American Journal of Human Genetics</i> , 2013, 93, 1158  Evaluation of common type 2 diabetes risk variants in a South Asian population of Sri Lankan descent. <i>PLoS ONE</i> , 2014, 9, e98608  Can surgeons assess CT suitability for endovascular repair (EVAR) in ruptured abdominal aortic aneurysm? Implications for a ruptured EVAR trial. <i>CardioVascular and Interventional Radiology</i> , 2008, 31, 865-9  Processes Underlying Glycemic Deterioration in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , 2021, 44, 511-518  Cohort Profile: East London Genes & Health (ELGH), a community based population genomics and health study of British-Bangladeshi and British-Pakistani people  Toppar: an interactive browser for viewing association study results. <i>Bioinformatics</i> , 2018, 34, 1922-192  Metabolic and reproductive characteristics of first-degree relatives of women with self-reported oligo-amenorrhoea and hirsutism. <i>Gynecological Endocrinology</i> , 2011, 27, 630-5  What will genome-wide association studies mean to the clinical endocrinologist?. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 2245-6  Variation at the IRF2 gene and susceptibility to psoriasis in chromosome 4q-linked families. <i>Journal of Investigative Dermatology</i> , 2004, 122, 640-3  Common maternal and fetal genetic variants show expected polygenic effects on risk of small- or large-for-gestational-age (SGA or LGA), except in the smallest 3% of babies. <i>PLoS Genetics</i> , 2020, 16, e1009191  Integrative network analysis highlights biological processes underlying GLP-1 stimulated insulin secretion: A DIRECT study. <i>PLoS ONE</i> , 2018, 13, e0189886  Analysis of overlapping genetic association in t	discovery and translation <i>Nature Genetics</i> , 2022,  A Multi-omic Integrative Scheme Characterizes Tissues of Action at Loci Associated with Type 2 Diabetes. <i>American Journal of Human Genetics</i> , 2020, 107, 1011-1028  Global Analysis of DNA Methylation Variation in Adipose Tissue from Twins Reveals Links to Disease-Associated Variants in Distal Regulatory Elements. <i>American Journal of Human Genetics</i> , 2013, 93, 1158  Evaluation of common type 2 diabetes risk variants in a South Asian population of Sri Lankan descent. <i>PLoS ONE</i> , 2014, 9, e98608  Can surgeons asses CT suitability for endovascular repair (EVAR) in ruptured abdominal aortic aneurysm? Implications for a ruptured EVAR trial. <i>CardioVascular and Interventional Radiology</i> , 2008, 21, 31, 865-9  Processes Underlying Glycemic Deterioration in Type 2 Diabetes: An IMI DIRECT Study. <i>Diabetes Care</i> , 2021, 44, 511-518  Cohort Profile: East London Genes & Health (ELGH), a community based population genomics and health study of British-Bangladeshi and British-Pakistani people  Toppar: an interactive browser for viewing association study results. <i>Bioinformatics</i> , 2018, 34, 1922-19247, 2  Metabolic and reproductive characteristics of first-degree relatives of women with self-reported oligo-amenorrhoea and hirsuitism. <i>Cynecological Endocrinology</i> , 2011, 27, 630-5  44  What will genome-wide association studies mean to the clinical endocrinologist?. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2009, 94, 224-5-6  Variation at the IRF2 gene and susceptibility to psoriasis in chromosome 4q-linked families. <i>Journal of Investigative Dermatology</i> , 2004, 122, 640-3  Common maternal and fetal genetic variants show expected polygenic effects on risk of small-or large-for-gestational-age (SGA or LGA), except in the smallest 3% of babies. <i>PLoS Genetics</i> , 2020, 61, 61, 61009191  Integrative network analysis highlights biological processes underlying GLP-1 stimulated insulin secretion: A DIRECT study. <i>PLoS ONE</i> , 2018, 13, e0189886  Analysis of overlapping genetic

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53	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes		4
52	Developing a network view of type 2 diabetes risk pathways through integration of genetic, genomic and functional data		4
51	Identification of type 2 diabetes loci in 433,540 East Asian individuals		4
50	Profiles of Glucose Metabolism in Different Prediabetes Phenotypes, Classified by Fasting Glycemia, 2-Hour OGTT, Glycated Hemoglobin, and 1-Hour OGTT: An IMI DIRECT Study. <i>Diabetes</i> , <b>2021</b> , 70, 2092-2106	0.9	4
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47	Genome-Wide Association Study of Peripheral Artery Disease. <i>Circulation Genomic and Precision Medicine</i> , <b>2021</b> , 14, e002862	5.2	3
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45	Whole blood co-expression modules associate with metabolic traits and type 2 diabetes: an IMI-DIRECT study. <i>Genome Medicine</i> , <b>2020</b> , 12, 109	14.4	3
44	Mendelian randomization analysis does not support causal associations of birth weight with hypertension risk and blood pressure in adulthood. <i>European Journal of Epidemiology</i> , <b>2020</b> , 35, 685-697	7 <sup>12.1</sup>	2
43	Genome-Wide Association Studies of Obesity <b>2014</b> , 33-53		2
42	Type 1 and type 2 diabetes-chalk and cheese?. <i>Diabetologia</i> , <b>2009</b> , 52, 1983-6	10.3	2
41	GANESH: software for customized annotation of genome regions. <i>Genome Research</i> , <b>2003</b> , 13, 2195-202	29.7	2
40	Polygenic Prediction of Type 2 Diabetes in Africa <i>Diabetes Care</i> , <b>2022</b> ,	14.6	2
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38	Aetiological differences between novel subtypes of diabetes derived from genetic associations		2
37	Rare Non-coding Variation Identified by Large Scale Whole Genome Sequencing Reveals Unexplained Heritability of Type 2 Diabetes		2
36	Genetic discovery and translational decision support from exome sequencing of 20,791 type 2 diabetes cases and 24,440 controls from five ancestries		2
35	Genome-wide scan and fine-mapping of rare nonsynonymous associations implicates intracellular lipolysis genes in fat distribution and cardio-metabolic risk		2
34	Causal analyses, statistical efficiency and phenotypic precision through Recall-by-Genotype study design	gn	2
33	Integration of human pancreatic islet genomic data refines regulatory mechanisms at Type 2 Diabetes susceptibility loci		2
32	Dietary metabolite profiling brings new insight into the relationship between nutrition and metabolic risk: An IMI DIRECT study. <i>EBioMedicine</i> , <b>2020</b> , 58, 102932	8.8	2
31	Large-Scale Analyses Provide No Evidence for Gene-Gene Interactions Influencing Type 2 Diabetes Risk. <i>Diabetes</i> , <b>2020</b> , 69, 2518-2522	0.9	2
30	HIV infection and anaemia do not affect HbA for the detection of diabetes in black South Africans: Evidence from the Durban Diabetes Study. <i>Diabetic Medicine</i> , <b>2021</b> , 38, e14605	3.5	2
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28	Genome-Wide Association Analysis of Pancreatic Beta-Cell Glucose Sensitivity. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2021</b> , 106, 80-90	5.6	2
27	Polygenic prediction of type 2 diabetes in continental Africa		2
26	Identification of genetic effects underlying type 2 diabetes in South Asian and European populations <i>Communications Biology</i> , <b>2022</b> , 5, 329	6.7	2
25	Multifaceted genome-wide study identifies novel regulatory loci in SLC22A11 and ZNF45 for body mass index in Indians. <i>Molecular Genetics and Genomics</i> , <b>2020</b> , 295, 1013-1026	3.1	1
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23	Four groups of type 2 diabetes contribute to the etiological and clinical heterogeneity in newly diagnosed individuals: An IMI DIRECT study <i>Cell Reports Medicine</i> , <b>2022</b> , 3, 100477	18	1
22	Analysis of overlapping genetic association in type 1 and type 2 diabetes		1
21	Determinants of penetrance and variable expressivity in monogenic metabolic conditions across 77,184 exomes		1

20	Genetic Predisposition to Coronary Artery Disease in Type 2 Diabetes Mellitus. <i>Circulation Genomic and Precision Medicine</i> , <b>2020</b> , 13, e002769	5.2	1
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11	Type 2 diabetes mellitus344-358		
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#### LIST OF PUBLICATIONS

- Homogeneity in the association of body mass index with type 2 diabetes across the UK Biobank: A Mendelian randomization study **2019**, 16, e1002982
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