Andrew Hattersley

List of Publications by Citations

709 papers	78,372 citations	133 h-index	263 g-index
747 ext. papers	90,533 ext. citations	9.6 avg, IF	7.26 L-index

#	Paper	IF	Citations
709	A common variant in the FTO gene is associated with body mass index and predisposes to childhood and adult obesity. *Science*, **2007**, 316, 889-94	33.3	3294
708	Genetic studies of body mass index yield new insights for obesity biology. *Nature*, **2015**, 518, 197-206	50.4	2687
707	Association analyses of 249,796 individuals reveal 18 new loci associated with body mass index. *Nature Genetics*, **2010**, 42, 937-48	36.3	2267
706	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes. *Science*, **2007**, 316, 1336-41	33.3	1823
705	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. *Nature Genetics*, **2010**, 42, 105-16	36.3	1673
704	Hundreds of variants clustered in genomic loci and biological pathways affect human height. *Nature*, **2010**, 467, 832-8	50.4	1514
703	Meta-analysis of genome-wide association data and large-scale replication identifies additional susceptibility loci for type 2 diabetes. *Nature Genetics*, **2008**, 40, 638-45	36.3	1496
702	Large-scale association analysis provides insights into the genetic architecture and pathophysiology of type 2 diabetes. *Nature Genetics*, **2012**, 44, 981-90	36.3	1482
701	Twelve type 2 diabetes susceptibility loci identified through large-scale association analysis. *Nature Genetics*, **2010**, 42, 579-89	36.3	1449
700	A reference panel of 64,976 haplotypes for genotype imputation. *Nature Genetics*, **2016**, 48, 1279-83	36.3	1447
699	Six new loci associated with body mass index highlight a neuronal influence on body weight regulation. *Nature Genetics*, **2009**, 41, 25-34	36.3	1368
698	Defining the role of common variation in the genomic and biological architecture of adult human height. *Nature Genetics*, **2014**, 46, 1173-86	36.3	1339
697	Association scan of 14,500 nonsynonymous SNPs in four diseases identifies autoimmunity variants. *Nature Genetics*, **2007**, 39, 1329-37	36.3	1130
696	Common variants near MC4R are associated with fat mass, weight and risk of obesity. *Nature Genetics*, **2008**, 40, 768-75	36.3	1048
695	Mutations in the hepatocyte nuclear factor-1alpha gene in maturity-onset diabetes of the young (MODY3). *Nature*, **1996**, 384, 455-8	50.4	973
694	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. *New England Journal of Medicine*, **2004**, 350, 1838-49	59.2	930
693	New genetic loci link adipose and insulin biology to body fat distribution. *Nature*, **2015**, 518, 187-196	50.4	920

692	Genome-wide trans-ancestry meta-analysis provides insight into the genetic architecture of type 2 diabetes susceptibility. *Nature Genetics*, **2014**, 46, 234-44	36.3	784
691	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. *New England Journal of Medicine*, **2006**, 355, 467-77	59.2	740
690	The fetal insulin hypothesis: an alternative explanation of the association of low birthweight with diabetes and vascular disease. *Lancet, The*, **1999**, 353, 1789-92	40	729
689	Meta-analysis identifies 13 new loci associated with waist-hip ratio and reveals sexual dimorphism in the genetic basis of fat distribution. *Nature Genetics*, **2010**, 42, 949-60	36.3	724
688	The genetic architecture of type 2 diabetes. *Nature*, **2016**, 536, 41-47	50.4	704
687	Fine-mapping type 2 diabetes loci to single-variant resolution using high-density imputation and islet-specific epigenome maps. *Nature Genetics*, **2018**, 50, 1505-1513	36.3	675
686	Genome-wide association analysis identifies 20 loci that influence adult height. *Nature Genetics*, **2008**, 40, 575-83	36.3	654
685	Genome-wide association study of CNVs in 16,000 cases of eight common diseases and 3,000 shared controls. *Nature*, **2010**, 464, 713-20	50.4	639
684	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. *Diabetes*, **2003**, 52, 568-72	0.9	614
683	Genetic variation in GIPR influences the glucose and insulin responses to an oral glucose challenge. *Nature Genetics*, **2010**, 42, 142-8	36.3	527
682	Mutations in the glucokinase gene of the fetus result in reduced birth weight. *Nature Genetics*, **1998**, 19, 268-70	36.3	483
681	Genetic cause of hyperglycaemia and response to treatment in diabetes. *Lancet, The*, **2003**, 362, 1275-81	40	437
680	Maturity-onset diabetes of the young (MODY): how many cases are we missing?. *Diabetologia*, **2010**, 53, 2504-8	10.3	432
679	Insulin gene mutations as a cause of permanent neonatal diabetes. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 15040-4	11.5	426
678	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. *Nature Genetics*, **2008**, 40, 949-51	36.3	417
677	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. *Diabetes*, **2017**, 66, 2888-2902	32	414
676	Rare and low-frequency coding variants alter human adult height. *Nature*, **2017**, 542, 186-190	50.4	412
675	What makes a good genetic association study?. *Lancet, The*, **2005**, 366, 1315-23	40	408

674	Genome-wide association scan meta-analysis identifies three Loci influencing adiposity and fat distribution. *PLoS Genetics*, **2009**, 5, e1000508	6	393
673	Clinical implications of a molecular genetic classification of monogenic beta-cell diabetes. *Nature Clinical Practice Endocrinology and Metabolism*, **2008**, 4, 200-13		367
672	Mutations in PTF1A cause pancreatic and cerebellar agenesis. *Nature Genetics*, **2004**, 36, 1301-5	36.3	356
671	Activating mutations in Kir6.2 and neonatal diabetes: new clinical syndromes, new scientific insights, and new therapy. *Diabetes*, **2005**, 54, 2503-13	0.9	356
670	Bayesian refinement of association signals for 14 loci in 3 common diseases. *Nature Genetics*, **2012**, 44, 1294-301	36.3	347
669	Linkage of type 2 diabetes to the glucokinase gene. *Lancet, The*, **1992**, 339, 1307-10	40	343
668	A common variant of HMGA2 is associated with adult and childhood height in the general population. *Nature Genetics*, **2007**, 39, 1245-50	36.3	330
667	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. *Nature Genetics*, **2014**, 46, 812-814	36.3	328
666	Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45,891 individuals. *PLoS Genetics*, **2012**, 8, e1002607	6	326
665	Concept, design and implementation of a cardiovascular gene-centric 50 k SNP array for large-scale genomic association studies. *PLoS ONE*, **2008**, 3, e3583	3.7	321
664	Common variants near ATM are associated with glycemic response to metformin in type 2 diabetes. *Nature Genetics*, **2011**, 43, 117-20	36.3	319
663	Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. *Diabetes*, **2004**, 53, 2713-8	0.9	314
662	Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. *Diabetologia*, **2008**, 51, 546-53	10.3	311
661	Mutations in the human delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. *Nature Genetics*, **2000**, 24, 438-41	36.3	308
660	Insulin mutation screening in 1,044 patients with diabetes: mutations in the INS gene are a common cause of neonatal diabetes but a rare cause of diabetes diagnosed in childhood or adulthood. *Diabetes*, **2008**, 57, 1034-42	0.9	299
659	Common variants in WFS1 confer risk of type 2 diabetes. *Nature Genetics*, **2007**, 39, 951-3	36.3	296
658	The clinical utility of C-peptide measurement in the care of patients with diabetes. *Diabetic Medicine*, **2013**, 30, 803-17	3.5	295
657	Genetic fine mapping and genomic annotation defines causal mechanisms at type 2 diabetes susceptibility loci. *Nature Genetics*, **2015**, 47, 1415-25	36.3	292

656	Macrosomia and hyperinsulinaemic hypoglycaemia in patients with heterozygous mutations in the HNF4A gene. *PLoS Medicine*, **2007**, 4, e118	11.6	279
655	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. *Diabetes*, **2007**, 56, 1930-7	0.9	273
654	Excess mortality and cardiovascular disease in young adults with type 1 diabetes in relation to age at onset: a nationwide, register-based cohort study. *Lancet, The*, **2018**, 392, 477-486	40	271
653	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. *American Journal of Human Genetics*, **2001**, 69, 553-69	11	271
652	C-reactive protein and its role in metabolic syndrome: mendelian randomisation study. *Lancet, The*, **2005**, 366, 1954-9	40	266
651	Genome-wide associations for birth weight and correlations with adult disease. *Nature*, **2016**, 538, 248-252	36.4	266
650	Common variation in the FTO gene alters diabetes-related metabolic traits to the extent expected given its effect on BMI. *Diabetes*, **2008**, 57, 1419-26	0.9	260
649	Meta-analysis of genome-wide association studies for body fat distribution in 694 649 individuals of European ancestry. *Human Molecular Genetics*, **2019**, 28, 166-174	5.6	258
648	Rfx6 directs islet formation and insulin production in mice and humans. *Nature*, **2010**, 463, 775-80	50.4	254
647	Variation in TCF7L2 influences therapeutic response to sulfonylureas: a GoDARTs study. *Diabetes*, **2007**, 56, 2178-82	0.9	251
646	Assessing the combined impact of 18 common genetic variants of modest effect sizes on type 2 diabetes risk. *Diabetes*, **2008**, 57, 3129-35	0.9	245
645	Regulation of Fto/Ftm gene expression in mice and humans. *American Journal of Physiology - Regulatory Integrative and Comparative Physiology*, **2008**, 294, R1185-96	3.2	238
644	Mutations in hepatocyte nuclear factor-1beta and their related phenotypes. *Journal of Medical Genetics*, **2006**, 43, 84-90	5.8	236
643	New loci associated with birth weight identify genetic links between intrauterine growth and adult height and metabolism. *Nature Genetics*, **2013**, 45, 76-82	36.3	232
642	Mutations in the hepatocyte nuclear factor-1beta gene are associated with familial hypoplastic glomerulocystic kidney disease. *American Journal of Human Genetics*, **2001**, 68, 219-24	11	227
641	Common variation in the DIO2 gene predicts baseline psychological well-being and response to combination thyroxine plus triiodothyronine therapy in hypothyroid patients. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 1623-9	5.6	224
640	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. *Diabetes*, **2006**, 55, 2640-4	0.9	222
639	Refining the accuracy of validated target identification through coding variant fine-mapping in type 2 diabetes. *Nature Genetics*, **2018**, 50, 559-571	36.3	221

638	The Influence of Age and Sex on Genetic Associations with Adult Body Size and Shape: A Large-Scale Genome-Wide Interaction Study. *PLoS Genetics*, **2015**, 11, e1005378	6	220
637	Prevalence, characteristics and clinical diagnosis of maturity onset diabetes of the young due to mutations in HNF1A, HNF4A, and glucokinase: results from the SEARCH for Diabetes in Youth. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 4055-62	5.6	219
636	Altered insulin secretory responses to glucose in diabetic and nondiabetic subjects with mutations in the diabetes susceptibility gene MODY3 on chromosome 12. *Diabetes*, **1996**, 45, 1503-10	0.9	216
635	Combining information from common type 2 diabetes risk polymorphisms improves disease prediction. *PLoS Medicine*, **2006**, 3, e374	11.6	214
634	The fat mass- and obesity-associated locus and dietary intake in children. *American Journal of Clinical Nutrition*, **2008**, 88, 971-8	7	213
633	The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatric Diabetes*, **2009**, 10 Suppl 12, 33-42	3.6	205
632	Molecular basis of Kir6.2 mutations associated with neonatal diabetes or neonatal diabetes plus neurological features. *Proceedings of the National Academy of Sciences of the United States of America*, **2004**, 101, 17539-44	11.5	205
631	Effective treatment with oral sulfonylureas in patients with diabetes due to sulfonylurea receptor 1 (SUR1) mutations. *Diabetes Care*, **2008**, 31, 204-9	14.6	203
630	Common variants of the novel type 2 diabetes genes CDKAL1 and HHEX/IDE are associated with decreased pancreatic beta-cell function. *Diabetes*, **2007**, 56, 3101-4	0.9	203
629	beta-cell genes and diabetes: molecular and clinical characterization of mutations in transcription factors. *Diabetes*, **2001**, 50 Suppl 1, S94-100	0.9	203
628	Improved genetic testing for monogenic diabetes using targeted next-generation sequencing. *Diabetologia*, **2013**, 56, 1958-63	10.3	201
627	Sensitivity to sulphonylureas in patients with hepatocyte nuclear factor-1alpha gene mutations: evidence for pharmacogenetics in diabetes. *Diabetic Medicine*, **2000**, 17, 543-5	3.5	198
626	The genetic abnormality in the beta cell determines the response to an oral glucose load. *Diabetologia*, **2002**, 45, 427-35	10.3	197
625	GATA6 haploinsufficiency causes pancreatic agenesis in humans. *Nature Genetics*, **2011**, 44, 20-22	36.3	195
624	Integrated genetic and epigenetic analysis identifies haplotype-specific methylation in the FTO type 2 diabetes and obesity susceptibility locus. *PLoS ONE*, **2010**, 5, e14040	3.7	193
623	Common variants in the TCF7L2 gene are strongly associated with type 2 diabetes mellitus in the Indian population. *Diabetologia*, **2007**, 50, 63-7	10.3	191
622	Frequency and phenotype of type 1 diabetes in the first six decades of life: a cross-sectional, genetically stratified survival analysis from UK Biobank. *Lancet Diabetes and Endocrinology,the*, **2018**, 6, 122-129	18.1	191
621	Mutations in KCNJ11, which encodes Kir6.2, are a common cause of diabetes diagnosed in the first 6 months of life, with the phenotype determined by genotype. *Diabetologia*, **2006**, 49, 1190-7	10.3	190

620	The majority of patients with long-duration type 1 diabetes are insulin microsecretors and have functioning beta cells. *Diabetologia*, **2014**, 57, 187-91	10.3	188
619	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. *Nature Genetics*, **2014**, 46, 61-64	36.3	187
618	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. *Lancet, The*, **2015**, 386, 957-63	40	186
617	Protein-altering variants associated with body mass index implicate pathways that control energy intake and expenditure in obesity. *Nature Genetics*, **2018**, 50, 26-41	36.3	186
616	Variants in ADCY5 and near CCNL1 are associated with fetal growth and birth weight. *Nature Genetics*, **2010**, 42, 430-5	36.3	184
615	Maturity-onset diabetes of the young: clinical heterogeneity explained by genetic heterogeneity. *Diabetic Medicine*, **1998**, 15, 15-24	3.5	184
614	Maternal and fetal genetic effects on birth weight and their relevance to cardio-metabolic risk factors. *Nature Genetics*, **2019**, 51, 804-814	36.3	181
613	Prevalence of vascular complications among patients with glucokinase mutations and prolonged, mild hyperglycemia. *JAMA - Journal of the American Medical Association*, **2014**, 311, 279-86	27.4	181
612	Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. *Diabetic Medicine*, **2008**, 25, 383-99	3.5	178
611	Missense mutations in the insulin promoter factor-1 gene predispose to type 2 diabetes. *Journal of Clinical Investigation*, **1999**, 104, R33-9	15.9	176
610	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. *Human Molecular Genetics*, **2006**, 15, 1793-800	5.6	175
609	The development and validation of a clinical prediction model to determine the probability of MODY in patients with young-onset diabetes. *Diabetologia*, **2012**, 55, 1265-72	10.3	172
608	Molecular genetics and phenotypic characteristics of MODY caused by hepatocyte nuclear factor 4alpha mutations in a large European collection. *Diabetologia*, **2005**, 48, 878-85	10.3	170
607	Definition, epidemiology and classification of diabetes in children and adolescents. *Pediatric Diabetes*, **2009**, 10 Suppl 12, 3-12	3.6	169
606	A genetic diagnosis of HNF1A diabetes alters treatment and improves glycaemic control in the majority of insulin-treated patients. *Diabetic Medicine*, **2009**, 26, 437-41	3.5	168
605	Relapsing diabetes can result from moderately activating mutations in KCNJ11. *Human Molecular Genetics*, **2005**, 14, 925-34	5.6	165
604	HNF1B-associated renal and extra-renal disease-an expanding clinical spectrum. *Nature Reviews Nephrology*, **2015**, 11, 102-12	14.9	163
603	Precision diabetes: learning from monogenic diabetes. *Diabetologia*, **2017**, 60, 769-777	10.3	162

602	Stratifying type 2 diabetes cases by BMI identifies genetic risk variants in LAMA1 and enrichment for risk variants in lean compared to obese cases. *PLoS Genetics*, **2012**, 8, e1002741	6	162
601	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. *American Journal of Human Genetics*, **2007**, 81, 375-82	11	161
600	Evaluation of common variants in the six known maturity-onset diabetes of the young (MODY) genes for association with type 2 diabetes. *Diabetes*, **2007**, 56, 685-93	0.9	160
599	Analysis of parent-offspring trios provides evidence for linkage and association between the insulin gene and type 2 diabetes mediated exclusively through paternally transmitted class III variable number tandem repeat alleles. *Diabetes*, **2000**, 49, 126-30	0.9	159
598	Kir6.2 mutations are a common cause of permanent neonatal diabetes in a large cohort of French patients. *Diabetes*, **2004**, 53, 2719-22	0.9	158
597	A distant upstream promoter of the HNF-4alpha gene connects the transcription factors involved in maturity-onset diabetes of the young. *Human Molecular Genetics*, **2001**, 10, 2089-97	5.6	158
596	A high prevalence of glucokinase mutations in gestational diabetic subjects selected by clinical criteria. *Diabetologia*, **2000**, 43, 250-3	10.3	158
595	Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation. *Diabetes Care*, **2015**, 38, 1383-92	14.6	157
594	Meta-analysis and a large association study confirm a role for calpain-10 variation in type 2 diabetes susceptibility. *American Journal of Human Genetics*, **2003**, 73, 1208-12	11	155
593	Studies of association between the gene for calpain-10 and type 2 diabetes mellitus in the United Kingdom. *American Journal of Human Genetics*, **2001**, 69, 544-52	11	154
592	Common nonsynonymous substitutions in SLCO1B1 predispose to statin intolerance in routinely treated individuals with type 2 diabetes: a go-DARTS study. *Clinical Pharmacology and Therapeutics*, **2011**, 89, 210-6	6.1	153
591	Genetic evidence that raised sex hormone binding globulin (SHBG) levels reduce the risk of type 2 diabetes. *Human Molecular Genetics*, **2010**, 19, 535-44	5.6	150
590	Genetic Evidence for Causal Relationships Between Maternal Obesity-Related Traits and Birth Weight. *JAMA - Journal of the American Medical Association*, **2016**, 315, 1129-40	27.4	149
589	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 3105-10	11.5	149
588	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. *Nature Communications*, **2015**, 6, 5897	17.4	147
587	A meta-analysis of thyroid-related traits reveals novel loci and gender-specific differences in the regulation of thyroid function. *PLoS Genetics*, **2013**, 9, e1003266	6	146
586	FTO gene variants are strongly associated with type 2 diabetes in South Asian Indians. *Diabetologia*, **2009**, 52, 247-52	10.3	145
585	A Type 1 Diabetes Genetic Risk Score Can Aid Discrimination Between Type 1 and Type 2 Diabetes in Young Adults. *Diabetes Care*, **2016**, 39, 337-44	14.6	141

584	Exploring the developmental overnutrition hypothesis using parental-offspring associations and FTO as an instrumental variable. *PLoS Medicine*, **2008**, 5, e33	11.6	139
583	A genome-wide association search for type 2 diabetes genes in African Americans. *PLoS ONE*, **2012**, 7, e29202	3.7	138
582	Mutations in the hepatocyte nuclear factor-1alpha gene are a common cause of maturity-onset diabetes of the young in the U.K. *Diabetes*, **1997**, 46, 720-5	0.9	138
581	Different genes, different diabetes: lessons from maturity-onset diabetes of the young. *Annals of Medicine*, **2002**, 34, 207-216	1.5	138
580	First UK survey of paediatric type 2 diabetes and MODY. *Archives of Disease in Childhood*, **2004**, 89, 526-9	2.2	137
579	ISPAD Clinical Practice Consensus Guidelines 2018: The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatric Diabetes*, **2018**, 19 Suppl 27, 47-63	3.6	136
578	ISPAD Clinical Practice Consensus Guidelines 2014. The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatric Diabetes*, **2014**, 15 Suppl 20, 47-64	3.6	133
577	Insights into the biochemical and genetic basis of glucokinase activation from naturally occurring hypoglycemia mutations. *Diabetes*, **2003**, 52, 2433-40	0.9	133
576	Reduced-function SLC22A1 polymorphisms encoding organic cation transporter 1 and glycemic response to metformin: a GoDARTS study. *Diabetes*, **2009**, 58, 1434-9	0.9	132
575	Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1 beta. *Kidney International*, **2000**, 57, 898-907	9.9	132
574	Missense glucokinase mutation in maturity-onset diabetes of the young and mutation screening in late-onset diabetes. *Nature Genetics*, **1992**, 2, 153-6	36.3	131
573	Disease progression and treatment response in data-driven subgroups of type 2 diabetes compared with models based on simple clinical features: an analysis using clinical trial data. *Lancet Diabetes and Endocrinology,the*, **2019**, 7, 442-451	18.1	128
572	Islet autoantibodies can discriminate maturity-onset diabetes of the young (MODY) from Type 1 diabetes. *Diabetic Medicine*, **2011**, 28, 1028-33	3.5	127
571	Loss-of-function CYP2C9 variants improve therapeutic response to sulfonylureas in type 2 diabetes: a Go-DARTS study. *Clinical Pharmacology and Therapeutics*, **2010**, 87, 52-6	6.1	124
570	Hepatocyte nuclear factor-1 beta mutations cause neonatal diabetes and intrauterine growth retardation: support for a critical role of HNF-1beta in human pancreatic development. *Diabetic Medicine*, **2006**, 23, 1301-6	3.5	123
569	A putative functional polymorphism in the IGF-I gene: association studies with type 2 diabetes, adult height, glucose tolerance, and fetal growth in U.K. populations. *Diabetes*, **2002**, 51, 2313-6	0.9	123
568	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. *PLoS Genetics*, **2014**, 10, e1004123	6	122
567	Cross-sectional and longitudinal studies suggest pharmacological treatment used in patients with glucokinase mutations does not alter glycaemia. *Diabetologia*, **2014**, 57, 54-6	10.3	122

566	A common genetic variant in the 15q24 nicotinic acetylcholine receptor gene cluster (CHRNA5-CHRNA3-CHRNB4) is associated with a reduced ability of women to quit smoking in pregnancy. *Human Molecular Genetics*, **2009**, 18, 2922-7	5.6	122
565	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. *Nature Genetics*, **2013**, 45, 947-50	36.3	120
564	Atypical familial juvenile hyperuricemic nephropathy associated with a hepatocyte nuclear factor-1beta gene mutation. *Kidney International*, **2003**, 63, 1645-51	9.9	120
563	KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes. *Human Mutation*, **2005**, 25, 22-7	4.7	118
562	Systematic Population Screening, Using Biomarkers and Genetic Testing, Identifies 2.5% of the U.K. Pediatric Diabetes Population With Monogenic Diabetes. *Diabetes Care*, **2016**, 39, 1879-1888	14.6	117
561	Type 2 diabetes risk alleles are associated with reduced size at birth. *Diabetes*, **2009**, 58, 1428-33	0.9	117
560	Homozygous mutations in NEUROD1 are responsible for a novel syndrome of permanent neonatal diabetes and neurological abnormalities. *Diabetes*, **2010**, 59, 2326-31	0.9	116
559	Permanent Neonatal Diabetes and Enteric Anendocrinosis Associated With Biallelic Mutations in NEUROG3. *Diabetes*, **2011**, 60, 1349-53	0.9	114
558	Persistent hyperinsulinemic hypoglycemia and maturity-onset diabetes of the young due to heterozygous HNF4A mutations. *Diabetes*, **2008**, 57, 1659-63	0.9	113
557	KCNJ11 activating mutations are associated with developmental delay, epilepsy and neonatal diabetes syndrome and other neurological features. *European Journal of Human Genetics*, **2006**, 14, 824-30	5.3	110
556	ISPAD Clinical Practice Consensus Guidelines 2006-2007. The diagnosis and management of monogenic diabetes in children. *Pediatric Diabetes*, **2006**, 7, 352-60	3.6	108
555	Regulation of apolipoprotein M gene expression by MODY3 gene hepatocyte nuclear factor-1alpha: haploinsufficiency is associated with reduced serum apolipoprotein M levels. *Diabetes*, **2003**, 52, 2989-95	0.9	108
554	Heterozygous ABCC8 mutations are a cause of MODY. *Diabetologia*, **2012**, 55, 123-7	10.3	105
553	Improved motor development and good long-term glycaemic control with sulfonylurea treatment in a patient with the syndrome of intermediate developmental delay, early-onset generalised epilepsy and neonatal diabetes associated with the V59M mutation in the KCNJ11 gene. *Diabetologia*, **2006**, 49, 2550-3	10.3	105
552	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. *Diabetes*, **2016**, 65, 2094-2099	0.9	105
551	GATA6 mutations cause a broad phenotypic spectrum of diabetes from pancreatic agenesis to adult-onset diabetes without exocrine insufficiency. *Diabetes*, **2013**, 62, 993-7	0.9	104
550	Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1beta mutations. *Kidney International*, **2002**, 61, 1243-51	9.9	104
549	Wolcott-Rallison syndrome is the most common genetic cause of permanent neonatal diabetes in consanguineous families. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4162-70	5.6	103

548	A common haplotype of the glucokinase gene alters fasting glucose and birth weight: association in six studies and population-genetics analyses. *American Journal of Human Genetics*, **2006**, 79, 991-1001	11	103
547	Maturity-onset diabetes of the young: from clinical description to molecular genetic characterization. *Best Practice and Research in Clinical Endocrinology and Metabolism*, **2001**, 15, 309-23	6.5	103
546	beta-cell genes and diabetes: quantitative and qualitative differences in the pathophysiology of hepatic nuclear factor-1alpha and glucokinase mutations. *Diabetes*, **2001**, 50 Suppl 1, S101-7	0.9	103
545	Analysis of transcription factors key for mouse pancreatic development establishes NKX2-2 and MNX1 mutations as causes of neonatal diabetes in man. *Cell Metabolism*, **2014**, 19, 146-54	24.6	102
544	Exome sequencing-driven discovery of coding polymorphisms associated with common metabolic phenotypes. *Diabetologia*, **2013**, 56, 298-310	10.3	102
543	A common variation in deiodinase 1 gene DIO1 is associated with the relative levels of free thyroxine and triiodothyronine. *Journal of Clinical Endocrinology and Metabolism*, **2008**, 93, 3075-81	5.6	102
542	Minireview: pharmacogenetics and beyond: the interaction of therapeutic response, beta-cell physiology, and genetics in diabetes. *Endocrinology*, **2006**, 147, 2657-63	4.8	101
541	Type 2 diabetes TCF7L2 risk genotypes alter birth weight: a study of 24,053 individuals. *American Journal of Human Genetics*, **2007**, 80, 1150-61	11	100
540	Isomers of the TCF1 gene encoding hepatocyte nuclear factor-1 alpha show differential expression in the pancreas and define the relationship between mutation position and clinical phenotype in monogenic diabetes. *Human Molecular Genetics*, **2006**, 15, 2216-24	5.6	100
539	Contrasting diabetes phenotypes associated with hepatocyte nuclear factor-1alpha and -1beta mutations. *Diabetes Care*, **2004**, 27, 1102-7	14.6	99
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