# **Andrew Hattersley**

# List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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

#	Paper	IF	Citations
709	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
708	Response to Comment on Meek et al. Reappearance of C-Peptide During the Third Trimester in Type 1 Diabetes Pregnancy: Pancreatic Regeneration or Fetal Hyperinsulinism? Diabetes Care 2021;44:1826-1834 <i>Diabetes Care</i> , <b>2022</b> , 45, e43-e44	14.6	
707	Evaluation of Evidence for Pathogenicity Demonstrates that BLK, KLF11 and PAX4 Should not be Included in Diagnostic Testing for MODY <i>Diabetes</i> , <b>2022</b> ,	0.9	2
706	Understanding the pathogenesis of lean non-autoimmune diabetes in an African population with newly diagnosed diabetes <i>Diabetologia</i> , <b>2022</b> , 65, 675	10.3	1
705	Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. <i>Diabetologia</i> , <b>2022</b> , 65, 246-249	10.3	
704	Alternative pre-analytic sample handling techniques for glucose measurement in the absence of fluoride tubes in low resource settings <i>PLoS ONE</i> , <b>2022</b> , 17, e0264432	3.7	
703	An HNF1[truncation associated with maturity-onset diabetes of the young impairs pancreatic progenitor differentiation by antagonizing HNF1[function Cell Reports, 2022, 38, 110425	10.6	O
702	Congenital beta cell defects are not associated with markers of islet autoimmunity, even in the context of high genetic risk for type 1 diabetes <i>Diabetologia</i> , <b>2022</b> , 1	10.3	О
701	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
700	Heterogeneity in phenotype, disease progression and drug response in type 2 diabetes <i>Nature Medicine</i> , <b>2022</b> , 28, 982-988	50.5	3
699	Islet autoantibody positivity in an adult population with recently diagnosed diabetes in Uganda. <i>PLoS ONE</i> , <b>2022</b> , 17, e0268783	3.7	
698	The power of genetic diversity in genome-wide association studies of lipids. <i>Nature</i> , <b>2021</b> ,	50.4	24
697	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. <i>Diabetologia</i> , <b>2021</b> , 1	10.3	1
696	Estimating disease prevalence in large datasets using genetic risk scores. <i>Nature Communications</i> , <b>2021</b> , 12, 6441	17.4	1
695	Syndromic Monogenic Diabetes Genes Should be Tested in Patients With a Clinical Suspicion of MODY. <i>Diabetes</i> , <b>2021</b> ,	0.9	2
694	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
693	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. <i>Cell Reports</i> , <b>2021</b> , 35, 108981	10.6	4

## (2020-2021)

692	Associations between low HDL, sex and cardiovascular risk markers are substantially different in sub-Saharan Africa and the UK: analysis of four population studies. <i>BMJ Global Health</i> , <b>2021</b> , 6,	6.6	1
691	Latent Autoimmune Diabetes of Adults (LADA) Is Likely to Represent a Mixed Population of Autoimmune (Type 1) and Nonautoimmune (Type 2) Diabetes. <i>Diabetes Care</i> , <b>2021</b> , 44, 1243-1251	14.6	10
690	Monogenic Diabetes and Integrated Stress Response Genes Display Altered Gene Expression in Type 1 Diabetes. <i>Diabetes</i> , <b>2021</b> , 70, 1885-1897	0.9	1
689	Reappearance of C-Peptide During the Third Trimester of Pregnancy in Type 1 Diabetes: Pancreatic Regeneration or Fetal Hyperinsulinism?. <i>Diabetes Care</i> , <b>2021</b> , 44, 1826-1834	14.6	3
688	In celebration of a century with insulin - Update of insulin gene mutations in diabetes. <i>Molecular Metabolism</i> , <b>2021</b> , 52, 101280	8.8	8
687	Identification of GCK-maturity-onset diabetes of the young in cases of neonatal hyperglycemia: A case series and review of clinical features. <i>Pediatric Diabetes</i> , <b>2021</b> , 22, 876-881	3.6	2
686	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
685	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated Permanent Neonatal Diabetes. <i>Diabetes Care</i> , <b>2021</b> , 44, 35-42	14.6	7
684	Associations Between Systolic Interarm Differences in Blood Pressure and Cardiovascular Disease Outcomes and Mortality: Individual Participant Data Meta-Analysis, Development and Validation of a Prognostic Algorithm: The INTERPRESS-IPD Collaboration. <i>Hypertension</i> , <b>2021</b> , 77, 650-661	8.5	7
683	Type 2 Diabetes and COVID-19-Related Mortality in the Critical Care Setting: A National Cohort Study in England, March-July 2020. <i>Diabetes Care</i> , <b>2021</b> , 44, 50-57	14.6	57
682	More on STAT1 Gain of Function, Type 1 Diabetes, and JAK Inhibition. <i>New England Journal of Medicine</i> , <b>2021</b> , 384, 93	59.2	
681	Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum Stress. <i>Diabetes</i> , <b>2021</b> , 70, 1006-1018	0.9	13
680	Two decades since the fetal insulin hypothesis: what have we learned from genetics?. <i>Diabetologia</i> , <b>2021</b> , 64, 717-726	10.3	3
679	The disproportionate excess mortality risk of COVID-19 in younger people with diabetes warrants vaccination prioritisation. <i>Diabetologia</i> , <b>2021</b> , 64, 1184-1186	10.3	9
678	Choice of HbA1c threshold for identifying individuals at high risk of type 2 diabetes and implications for diabetes prevention programmes: a cohort study. <i>BMC Medicine</i> , <b>2021</b> , 19, 184	11.4	O
677	Higher maternal adiposity reduces offspring birthweight if associated with a metabolically favourable profile. <i>Diabetologia</i> , <b>2021</b> , 64, 2790-2802	10.3	0
676	Phantasia-The psychological significance of lifelong visual imagery vividness extremes. <i>Cortex</i> , <b>2020</b> , 130, 426-440	3.8	31
675	Noninvasive Fetal Genotyping by Droplet Digital PCR to Identify Maternally Inherited Monogenic Diabetes Variants. <i>Clinical Chemistry</i> , <b>2020</b> , 66, 958-965	5.5	13

674	Precision medicine in diabetes: a Consensus Report from the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). <i>Diabetologia</i> , <b>2020</b> , 63, 1671-1693	10.3	33
673	Precision Medicine in Diabetes: A Consensus Report From the American Diabetes Association (ADA) and the European Association for the Study of Diabetes (EASD). <i>Diabetes Care</i> , <b>2020</b> , 43, 1617-1635	14.6	75
672	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
671	Prior event rate ratio adjustment produced estimates consistent with randomized trial: a diabetes case study. <i>Journal of Clinical Epidemiology</i> , <b>2020</b> , 122, 78-86	5.7	5
670	Studies of insulin and proinsulin in pancreas and serum support the existence of aetiopathological endotypes of type 1 diabetes associated with age at diagnosis. <i>Diabetologia</i> , <b>2020</b> , 63, 1258-1267	10.3	40
669	The challenge of diagnosing type 1 diabetes in older adults. <i>Diabetic Medicine</i> , <b>2020</b> , 37, 1781-1782	3.5	2
668	The role of physical activity in metabolic homeostasis before and after the onset of type 2 diabetes: an IMI DIRECT study. <i>Diabetologia</i> , <b>2020</b> , 63, 744-756	10.3	4
667	Absence of Islet Autoantibodies and Modestly Raised Glucose Values at Diabetes Diagnosis Should Lead to Testing for MODY: Lessons From a 5-Year Pediatric Swedish National Cohort Study. <i>Diabetes Care</i> , <b>2020</b> , 43, 82-89	14.6	35
666	Risk factors for genital infections in people initiating SGLT2 inhibitors and their impact on discontinuation. <i>BMJ Open Diabetes Research and Care</i> , <b>2020</b> , 8,	4.5	14
665	TriMaster: randomised double-blind crossover study of a DPP4 inhibitor, SGLT2 inhibitor and thiazolidinedione as second-line or third-line therapy in patients with type 2 diabetes who have suboptimal glycaemic control on metformin treatment with or without a sulfonylurea-a	3	6
664	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. <i>Journal of Clinical Investigation</i> , <b>2020</b> , 130, 14-16	15.9	13
663	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, <b>2020</b> , 130, 6338-6353	15.9	21
662	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> , <b>2020</b> , 16, e1009191	6	5
661	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. <i>Wellcome Open Research</i> , <b>2020</b> , 5, 175	4.8	O
660	A Non-Coding Disease Modifier of Pancreatic Agenesis Identified by Genetic Correction in a Patient-Derived iPSC Line. <i>Cell Stem Cell</i> , <b>2020</b> , 27, 137-146.e6	18	8
659	Identifying routine clinical predictors of non-adherence to second-line therapies in type 2 diabetes: A retrospective cohort analysis in a large primary care database. <i>Diabetes, Obesity and Metabolism</i> , <b>2020</b> , 22, 59-65	6.7	6
658	De Novo Mutations in Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. <i>Diabetes</i> , <b>2020</b> , 69, 477-483	0.9	17
657	Type 1 diabetes can present before the age of 6[months and is characterised by autoimmunity and rapid loss of beta cells. <i>Diabetologia</i> , <b>2020</b> , 63, 2605-2615	10.3	9

656	Novel loci for childhood body mass index and shared heritability with adult cardiometabolic traits. <i>PLoS Genetics</i> , <b>2020</b> , 16, e1008718	6	25
655	Predicting post one-year durability of glucose-lowering monotherapies in patients with newly-diagnosed type 2 diabetes mellitus - A MASTERMIND precision medicine approach (UKPDS 87). <i>Diabetes Research and Clinical Practice</i> , <b>2020</b> , 166, 108333	7.4	2
654	Monogenic Diabetes: From Genetic Insights to Population-Based Precision in Care. Reflections From a EditorsNExpert Forum. <i>Diabetes Care</i> , <b>2020</b> , 43, 3117-3128	14.6	23
653	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
652	Risk of Anemia With Metformin Use in Type 2 Diabetes: A MASTERMIND Study. <i>Diabetes Care</i> , <b>2020</b> , 43, 2493-2499	14.6	10
651	Clinical Characteristics and Long-term Follow-up of Patients with Diabetes Due To PTF1A Enhancer Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2020</b> , 105,	5.6	6
650	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. <i>Wellcome Open Research</i> , <b>2020</b> , 5, 175	4.8	0
649	Meta-analysis of up to 622,409 individuals identifies 40 novel smoking behaviour associated genetic loci. <i>Molecular Psychiatry</i> , <b>2020</b> , 25, 2392-2409	15.1	45
648	A Mendelian Randomization Study Provides Evidence That Adiposity and Dyslipidemia Lead to Lower Urinary Albumin-to-Creatinine Ratio, a Marker of Microvascular Function. <i>Diabetes</i> , <b>2020</b> , 69, 10	72 <sup>2</sup> 108	<b>2</b> 7
647	Strategies to identify individuals with monogenic diabetes: results of an economic evaluation. <i>BMJ Open</i> , <b>2020</b> , 10, e034716	3	5
647		3	5
	Open, <b>2020</b> , 10, e034716  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and	3	5
646	Open, 2020, 10, e034716  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and	3	5
646	Open, 2020, 10, e034716  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and	3	5
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646 645 644	Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149	8	16
646 645 644 642	Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  Predicting and elucidating the etiology of fatty liver disease: A machine learning modeling and validation study in the IMI DIRECT cohorts 2020, 17, e1003149  GATA6 Cooperates with EOMES/SMAD2/3 to Deploy the Gene Regulatory Network Governing		

638	Clusters provide a better holistic view of type 2 diabetes than simple clinical features - AuthorsN reply. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2019</b> , 7, 669	18.1	1
637	Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 275-286	11	80
636	Discovery of biomarkers for glycaemic deterioration before and after the onset of type 2 diabetes: descriptive characteristics of the epidemiological studies within the IMI DIRECT Consortium. <i>Diabetologia</i> , <b>2019</b> , 62, 1601-1615	10.3	14
635	Association of maternal circulating 25(OH)D and calcium with birth weight: A mendelian randomisation analysis. <i>PLoS Medicine</i> , <b>2019</b> , 16, e1002828	11.6	20
634	Genetic studies of abdominal MRI data identify genes regulating hepcidin as major determinants of liver iron concentration. <i>Journal of Hepatology</i> , <b>2019</b> , 71, 594-602	13.4	10
633	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 985-989	11	28
632	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. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2019</b> , 7, 442-451	18.1	128
631	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
630	Time trends in prescribing of type 2 diabetes drugs, glycaemic response and risk factors: A retrospective analysis of primary care data, 2010-2017. <i>Diabetes, Obesity and Metabolism</i> , <b>2019</b> , 21, 157	6 <sup>6</sup> 7584	, 34
629	Understanding the manifestation of diabetes in sub Saharan Africa to inform therapeutic approaches and preventive strategies: a narrative review. <i>Clinical Diabetes and Endocrinology</i> , <b>2019</b> , 5, 2	4.7	26
628	Persistent C-peptide is associated with reduced hypoglycaemia but not HbA in adults with longstanding Type 1 diabetes: evidence for lack of intensive treatment in UK clinical practice?. <i>Diabetic Medicine</i> , <b>2019</b> , 36, 1092-1099	3.5	21
627	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. <i>Diabetes</i> , <b>2019</b> , 68, 1528-1535	0.9	15
626	Type 1 diabetes defined by severe insulin deficiency occurs after 30 lyears of age and is commonly treated as type 2 diabetes. <i>Diabetologia</i> , <b>2019</b> , 62, 1167-1172	10.3	48
625	What to do with diabetes therapies when HbA1c lowering is inadequate: add, switch, or continue? A MASTERMIND study. <i>BMC Medicine</i> , <b>2019</b> , 17, 79	11.4	6
624	Type 1 diabetes genetic risk score discriminates between monogenic and Type 1 diabetes in children diagnosed at the age of . <i>Diabetic Medicine</i> , <b>2019</b> , 36, 1694-1702	3.5	8
623	A genome-wide association study implicates multiple mechanisms influencing raised urinary albumin-creatinine ratio. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 4197-4207	5.6	8
622	Protein-coding variants implicate novel genes related to lipid homeostasis contributing to body-fat distribution. <i>Nature Genetics</i> , <b>2019</b> , 51, 452-469	36.3	44
621	Patterns of postmeal insulin secretion in individuals with sulfonylurea-treated neonatal diabetes show predominance of non-K-channel pathways. <i>BMJ Open Diabetes Research and Care</i> , <b>2019</b> , 7, e00072	4.5	4

620	Development and validation of multivariable clinical diagnostic models to identify type 1 diabetes requiring rapid insulin therapy in adults aged 18-50 years. <i>BMJ Open</i> , <b>2019</b> , 9, e031586	3	19
619	Zinc Transporter 8 Autoantibodies (ZnT8A) and a Type 1 Diabetes Genetic Risk Score Can Exclude Individuals With Type 1 Diabetes From Inappropriate Genetic Testing for Monogenic Diabetes. <i>Diabetes Care</i> , <b>2019</b> , 42, e16-e17	14.6	15
618	Genome-Wide and Abdominal MRI Data Provide Evidence That a Genetically Determined Favorable Adiposity Phenotype Is Characterized by Lower Ectopic Liver Fat and Lower Risk of Type 2 Diabetes, Heart Disease, and Hypertension. <i>Diabetes</i> , <b>2019</b> , 68, 207-219	0.9	46
617	A Type 1 Diabetes Genetic Risk Score Can Identify Patients With GAD65 Autoantibody-Positive Type 2 Diabetes Who Rapidly Progress to Insulin Therapy. <i>Diabetes Care</i> , <b>2019</b> , 42, 208-214	14.6	20
616	Cognitive, Neurological, and Behavioral Features in Adults With Neonatal Diabetes. <i>Diabetes Care</i> , <b>2019</b> , 42, 215-224	14.6	11
615	Meta-analysis of genome-wide association studies for body fat distribution in 694\(\bar{b}\)49 individuals of European ancestry. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 166-174	5.6	258
614	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 982-986	8.1	15
613	Fetal Genotype and Maternal Glucose Have Independent and Additive Effects on Birth Weight. <i>Diabetes</i> , <b>2018</b> , 67, 1024-1029	0.9	24
612	Genetic risk scores in adult-onset type 1 diabetes - AuthorsNeply. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2018</b> , 6, 169	18.1	3
611	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	5.6	98
610	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
609	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. <i>Diabetologia</i> , <b>2018</b> , 61, 1027-1036	10.3	17
608	Precision Medicine in Type 2 Diabetes: Clinical Markers of Insulin Resistance Are Associated With Altered Short- and Long-term Glycemic Response to DPP-4 Inhibitor Therapy. <i>Diabetes Care</i> , <b>2018</b> , 41, 705-712	14.6	36
607	A type 1 diabetes genetic risk score can discriminate monogenic autoimmunity with diabetes from early-onset clustering of polygenic autoimmunity with diabetes. <i>Diabetologia</i> , <b>2018</b> , 61, 862-869	10.3	20
606	Comment on Dubois-Laforgue et al. Diabetes, Associated Clinical Spectrum, Long-term Prognosis, and Genotype/Phenotype Correlations in 201 Adult Patients With Hepatocyte Nuclear Factor 1B () Molecular Defects. Diabetes Care 2017;40:1436-1443. <i>Diabetes Care</i> , <b>2018</b> , 41, e7	14.6	3
605	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. <i>Journal of Pediatric Endocrinology and Metabolism</i> , <b>2018</b> , 31, 195-204	1.6	8
604	Prevalence of diabetes in Australia: insights from the Fremantle Diabetes Study Phase II. <i>Internal Medicine Journal</i> , <b>2018</b> , 48, 803-809	1.6	19
603	Are we missing hypoglycaemia? Elderly patients with insulin-treated diabetes present to primary care frequently with non-specific symptoms associated with hypoglycaemia. <i>Primary Care Diabetes</i> , <b>2018</b> , 12, 139-146	2.4	16

602	Maternal and fetal genetic contribution to gestational weight gain. <i>International Journal of Obesity</i> , <b>2018</b> , 42, 775-784	5.5	19
601	Genome-wide methylomic analysis in individuals with HNF1B intragenic mutation and 17q12 microdeletion. <i>Clinical Epigenetics</i> , <b>2018</b> , 10, 97	7.7	10
600	Sex and BMI Alter the Benefits and Risks of Sulfonylureas and Thiazolidinediones in Type 2 Diabetes: A Framework for Evaluating Stratification Using Routine Clinical and Individual Trial Data. <i>Diabetes Care</i> , <b>2018</b> , 41, 1844-1853	14.6	43
599	Time trends and geographical variation in prescribing of drugs for diabetes in England from 1998 to 2017. <i>Diabetes, Obesity and Metabolism</i> , <b>2018</b> , 20, 2159-2168	6.7	38
598	Molecular reductions in glucokinase activity increase counter-regulatory responses to hypoglycemia in mice and humans with diabetes. <i>Molecular Metabolism</i> , <b>2018</b> , 17, 17-27	8.8	25
597	Excess mortality and cardiovascular disease in young adults with type 1 diabetes in relation to age at onset: a nationwide, register-based cohort study. <i>Lancet, The</i> , <b>2018</b> , 392, 477-486	40	271
596	Exocrine pancreatic dysfunction is common in hepatocyte nuclear factor 1Eassociated renal disease and can be symptomatic. <i>CKJ: Clinical Kidney Journal</i> , <b>2018</b> , 11, 453-458	4.5	5
595	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2018</b> , 6, 637-646	18.1	77
594	C-Peptide Decline in Type 1 Diabetes Has Two Phases: An Initial Exponential Fall and a Subsequent Stable Phase. <i>Diabetes Care</i> , <b>2018</b> , 41, 1486-1492	14.6	54
593	The Common Variant I27L Is a Modifier of Age at Diabetes Diagnosis in Individuals With HNF1A-MODY. <i>Diabetes</i> , <b>2018</b> , 67, 1903-1907	0.9	7
592	Permanent neonatal diabetes: combining sulfonylureas with insulin may be an effective treatment. <i>Diabetic Medicine</i> , <b>2018</b> , 35, 1291	3.5	7
591	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
590	Frequency and phenotype of type 1 diabetes in the first six decades of life: a cross-sectional, genetically stratified survival analysis from UK Biobank. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2018</b> , 6, 122-129	18.1	191
589	Marked intrafamilial variability of exocrine and endocrine pancreatic phenotypes due to a splice site mutation in GATA6. <i>Biotechnology and Biotechnological Equipment</i> , <b>2018</b> , 32, 124-129	1.6	
588	Evaluating associations between the benefits and risks of drug therapy in type 2 diabetes: a joint modeling approach. <i>Clinical Epidemiology</i> , <b>2018</b> , 10, 1869-1877	5.9	8
587	Future Roadmaps for Precision Medicine Applied to Diabetes: Rising to the Challenge of Heterogeneity. <i>Journal of Diabetes Research</i> , <b>2018</b> , 2018, 3061620	3.9	11
586	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
585	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2018</b> , 103, 3225-3230	5.6	14

584	ISPAD Clinical Practice Consensus Guidelines 2018: The diagnosis and management of monogenic diabetes in children and adolescents. <i>Pediatric Diabetes</i> , <b>2018</b> , 19 Suppl 27, 47-63	3.6	136
583	A UK nationwide prospective study of treatment change in MODY: genetic subtype and clinical characteristics predict optimal glycaemic control after discontinuing insulin and metformin. <i>Diabetologia</i> , <b>2018</b> , 61, 2520-2527	10.3	38
582	Genetic scores to stratify risk of developing multiple islet autoantibodies and type 1 diabetes: A prospective study in children. <i>PLoS Medicine</i> , <b>2018</b> , 15, e1002548	11.6	60
581	Rare and low-frequency coding variants alter human adult height. <i>Nature</i> , <b>2017</b> , 542, 186-190	50.4	412
580	Hyperglycaemia-related complications at the time of diagnosis can cause permanent neurological disability in children with neonatal diabetes. <i>Diabetic Medicine</i> , <b>2017</b> , 34, 1000-1004	3.5	8
579	Dominant ER Stress-Inducing Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. <i>Diabetes</i> , <b>2017</b> , 66, 2044-2053	0.9	56
578	Recessively Inherited Mutations Cause Autoimmunity Presenting as Neonatal Diabetes. <i>Diabetes</i> , <b>2017</b> , 66, 2316-2322	0.9	39
577	Neuropsychological impairments in children with KCNJ11 neonatal diabetes. <i>Diabetic Medicine</i> , <b>2017</b> , 34, 1171-1173	3.5	10
576	A successful transition to sulfonylurea treatment in male infant with neonatal diabetes caused by the novel abcc8 gene mutation and three years follow-up. <i>Diabetes Research and Clinical Practice</i> , <b>2017</b> , 129, 59-61	7·4	5
575	Management of sulfonylurea-treated monogenic diabetes in pregnancy: implications of placental glibenclamide transfer. <i>Diabetic Medicine</i> , <b>2017</b> , 34, 1332-1339	3.5	21
574	An Expanded Genome-Wide Association Study of Type 2 Diabetes in Europeans. <i>Diabetes</i> , <b>2017</b> , 66, 288	38 <del>-</del> 2 <i>9</i> 02	2 414
573	A Low-Frequency Inactivating Variant Enriched in the Finnish Population Is Associated With Fasting Insulin Levels and Type 2 Diabetes Risk. <i>Diabetes</i> , <b>2017</b> , 66, 2019-2032	0.9	29
572	Precision diabetes: learning from monogenic diabetes. <i>Diabetologia</i> , <b>2017</b> , 60, 769-777	10.3	162
57 <sup>1</sup>	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. <i>Nature Communications</i> , <b>2017</b> , 8, 888	17.4	57
570	Cohort profile for the MASTERMIND study: using the Clinical Practice Research Datalink (CPRD) to investigate stratification of response to treatment in patients with type 2 diabetes. <i>BMJ Open</i> , <b>2017</b> , 7, e017989	3	13
569	The prevalence of monogenic diabetes in Australia: the Fremantle Diabetes Study Phase II. <i>Medical Journal of Australia</i> , <b>2017</b> , 207, 344-347	4	11
568	Costs and Treatment Pathways for Type 2 Diabetes in the UK: A Mastermind Cohort Study. <i>Diabetes Therapy</i> , <b>2017</b> , 8, 1031-1045	3.6	8
567	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. <i>Diabetologia</i> , <b>2017</b> , 60, 2168-2173	10.3	7

566	Population-Based Assessment of a Biomarker-Based Screening Pathway to Aid Diagnosis of Monogenic Diabetes in Young-Onset Patients. <i>Diabetes Care</i> , <b>2017</b> , 40, 1017-1025	14.6	73
565	Pancreas and gallbladder agenesis in a newborn with semilobar holoprosencephaly, a case report. <i>BMC Medical Genetics</i> , <b>2017</b> , 18, 57	2.1	3
564	Defining drug response for stratified medicine. <i>Drug Discovery Today</i> , <b>2017</b> , 22, 173-179	8.8	13
563	Analysis of cell-free fetal DNA for non-invasive prenatal diagnosis in a family with neonatal diabetes. <i>Diabetic Medicine</i> , <b>2017</b> , 34, 582-585	3.5	24
562	Sequence data and association statistics from 12,940 type 2 diabetes cases and controls. <i>Scientific Data</i> , <b>2017</b> , 4, 170179	8.2	22
561	Markers of ECell Failure Predict Poor Glycemic Response to GLP-1 Receptor Agonist Therapy in Type 2 Diabetes. <i>Diabetes Care</i> , <b>2016</b> , 39, 250-7	14.6	94
560	A reference panel of 64,976 haplotypes for genotype imputation. <i>Nature Genetics</i> , <b>2016</b> , 48, 1279-83	36.3	1447
559	Prematurity and Genetic Testing for Neonatal Diabetes. <i>Pediatrics</i> , <b>2016</b> , 138,	7.4	19
558	Hypogonadotropic Hypogonadism and Short Stature in Patients with Diabetes Due to Neurogenin 3 Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2016</b> , 101, 3555-3558	5.6	6
557	A principal component meta-analysis on multiple anthropometric traits identifies novel loci for body shape. <i>Nature Communications</i> , <b>2016</b> , 7, 13357	17.4	46
556	Beta cell function and ongoing autoimmunity in long-standing, childhood onset type 1 diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 2722-2726	10.3	33
555	Psychiatric morbidity in children with KCNJ11 neonatal diabetes. <i>Diabetic Medicine</i> , <b>2016</b> , 33, 1387-91	3.5	22
554	Practical Classification Guidelines for Diabetes in patients treated with insulin: a cross-sectional study of the accuracy of diabetes diagnosis. <i>British Journal of General Practice</i> , <b>2016</b> , 66, e315-22	1.6	35
553	Systematic Population Screening, Using Biomarkers and Genetic Testing, Identifies 2.5% of the U.K. Pediatric Diabetes Population With Monogenic Diabetes. <i>Diabetes Care</i> , <b>2016</b> , 39, 1879-1888	14.6	117
552	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. <i>Diabetes</i> , <b>2016</b> , 65, 2810-5	0.9	16
551	Maternal hypothyroxinaemia in pregnancy is associated with obesity and adverse maternal metabolic parameters. <i>European Journal of Endocrinology</i> , <b>2016</b> , 174, 51-7	6.5	39
550	Characteristics of maturity onset diabetes of the young in a large diabetes center. <i>Pediatric Diabetes</i> , <b>2016</b> , 17, 360-7	3.6	36
549	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

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548	A Type 1 Diabetes Genetic Risk Score Can Aid Discrimination Between Type 1 and Type 2 Diabetes in Young Adults. <i>Diabetes Care</i> , <b>2016</b> , 39, 337-44	14.6	141
547	Adherence to Oral Glucose-Lowering Therapies and Associations With 1-Year HbA1c: A Retrospective Cohort Analysis in a Large Primary Care Database. <i>Diabetes Care</i> , <b>2016</b> , 39, 258-263	14.6	57
546	Should Studies of Diabetes Treatment Stratification Correct for Baseline HbA1c?. <i>PLoS ONE</i> , <b>2016</b> , 11, e0152428	3.7	21
545	The genetic architecture of type 2 diabetes. <i>Nature</i> , <b>2016</b> , 536, 41-47	50.4	704
544	Crossover studies can help the individualisation of care in type 2 diabetes: the MASTERMIND approach. <i>Practical Diabetes</i> , <b>2016</b> , 33, 115-117	0.7	
543	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. <i>Kidney International</i> , <b>2016</b> , 90, 203-11	9.9	42
542	Random non-fasting C-peptide: bringing robust assessment of endogenous insulin secretion to the clinic. <i>Diabetic Medicine</i> , <b>2016</b> , 33, 1554-1558	3.5	34
541	Low IgE Is a Useful Tool to Identify STAT3 Gain-of-Function Mutations. Clinical Chemistry, <b>2016</b> , 62, 153	6 <del>-5</del> 1. <b>5</b> 38	4
540	South Asian individuals with diabetes who are referred for MODY testing in the UK have a lower mutation pick-up rate than white European people. <i>Diabetologia</i> , <b>2016</b> , 59, 2262-5	10.3	13
539	Monogenic Causes of Diabetes <b>2016</b> , 241-261		1
<ul><li>539</li><li>538</li></ul>	Monogenic Causes of Diabetes <b>2016</b> , 241-261  Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6	10.3	54
	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation	0.9	
538	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6  Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes.		54
538 537	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6  Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. <i>Diabetes</i> , <b>2016</b> , 65, 2094-2099  Differential regulation of serum microRNA expression by HNF1[and HNF1[aranscription factors.	0.9	54 105 15
538 537 536	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6  Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. <i>Diabetes</i> , <b>2016</b> , 65, 2094-2099  Differential regulation of serum microRNA expression by HNF1land HNF1ltranscription factors. <i>Diabetologia</i> , <b>2016</b> , 59, 1463-1473  Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in	0.9	54 105 15
<ul><li>538</li><li>537</li><li>536</li><li>535</li></ul>	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6  Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. <i>Diabetes</i> , <b>2016</b> , 65, 2094-2099  Differential regulation of serum microRNA expression by HNF1[and HNF1[transcription factors. <i>Diabetologia</i> , <b>2016</b> , 59, 1463-1473  Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in ATP sensitivity markedly affect diabetes risk. <i>Diabetologia</i> , <b>2016</b> , 59, 1430-1436  Genetic Evidence for a Link Between Favorable Adiposity and Lower Risk of Type 2 Diabetes,	0.9	<ul><li>54</li><li>105</li><li>15</li><li>18</li></ul>
<ul><li>538</li><li>537</li><li>536</li><li>535</li><li>534</li></ul>	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. <i>Diabetologia</i> , <b>2016</b> , 59, 1162-6  Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. <i>Diabetes</i> , <b>2016</b> , 65, 2094-2099  Differential regulation of serum microRNA expression by HNF1[and HNF1[branscription factors. <i>Diabetologia</i> , <b>2016</b> , 59, 1463-1473  Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in ATP sensitivity markedly affect diabetes risk. <i>Diabetologia</i> , <b>2016</b> , 59, 1430-1436  Genetic Evidence for a Link Between Favorable Adiposity and Lower Risk of Type 2 Diabetes, Hypertension, and Heart Disease. <i>Diabetes</i> , <b>2016</b> , 65, 2448-60	0.9	<ul><li>54</li><li>105</li><li>15</li><li>18</li><li>86</li></ul>

530	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. <i>Lancet, The</i> , <b>2015</b> , 386, 957-63	40	186
529	Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation. <i>Diabetes Care</i> , <b>2015</b> , 38, 1383-92	14.6	157
528	A Missense Mutation in PPP1R15B Causes a Syndrome Including Diabetes, Short Stature, and Microcephaly. <i>Diabetes</i> , <b>2015</b> , 64, 3951-62	0.9	48
527	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	0.9	20
526	Monogenic disorders of the Itell <b>2015</b> , 426-441		
525	Clinical characteristics and molecular genetic analysis of 22 patients with neonatal diabetes from the South-Eastern region of Turkey: predominance of non-KATP channel mutations. <i>European Journal of Endocrinology</i> , <b>2015</b> , 172, 697-705	6.5	40
524	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. <i>European Journal of Human Genetics</i> , <b>2015</b> , 23, 1744-8	5.3	24
523	Type 2 Diabetes, SGLT2 Inhibitors, and Glucose Secretion. <i>New England Journal of Medicine</i> , <b>2015</b> , 373, 974-6	59.2	27
522	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. <i>Journal of Pediatric Endocrinology and Metabolism</i> , <b>2015</b> , 28, 1279-86	1.6	25
521	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
520	HNF1B-associated renal and extra-renal disease-an expanding clinical spectrum. <i>Nature Reviews Nephrology</i> , <b>2015</b> , 11, 102-12	14.9	163
519	The value of in vitro studies in a case of neonatal diabetes with a novel Kir6.2-W68G mutation. <i>Clinical Case Reports (discontinued)</i> , <b>2015</b> , 3, 884-7	0.7	4
518	Can clinical features be used to differentiate type 1 from type 2 diabetes? A systematic review of the literature. <i>BMJ Open</i> , <b>2015</b> , 5, e009088	3	53
517	3. A Novel, Inexpensive Test Can Discriminate between Type 1 and Type 2 Diabetes (1745-P). <i>Nederlands Tijdschrift Voor Diabetologie</i> , <b>2015</b> , 13, 57-57	O	
516	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
515	Optimisation of an Advanced Oxidation Protein Products Assay: Its Application to Studies of Oxidative Stress in Diabetes Mellitus. <i>Oxidative Medicine and Cellular Longevity</i> , <b>2015</b> , 2015, 496271	6.7	29
514	Most people with long-duration type 1 diabetes in a large population-based study are insulin microsecretors. <i>Diabetes Care</i> , <b>2015</b> , 38, 323-8	14.6	76
513	Expanding the Clinical Spectrum Associated With GLIS3 Mutations. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2015</b> , 100, E1362-9	5.6	53

512	Assessment of the HNF1B Score as a Tool to Select Patients for HNF1B Genetic Testing. <i>Nephron</i> , <b>2015</b> , 130, 134-40	3.3	9
511	New genetic loci link adipose and insulin biology to body fat distribution. <i>Nature</i> , <b>2015</b> , 518, 187-196	50.4	920
510	Genetic studies of body mass index yield new insights for obesity biology. <i>Nature</i> , <b>2015</b> , 518, 197-206	50.4	2687
509	Low-frequency and rare exome chip variants associate with fasting glucose and type 2 diabetes susceptibility. <i>Nature Communications</i> , <b>2015</b> , 6, 5897	17.4	147
508	Lower Circulating B12 Is Associated with Higher Obesity and Insulin Resistance during Pregnancy in a Non-Diabetic White British Population. <i>PLoS ONE</i> , <b>2015</b> , 10, e0135268	3.7	54
507	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. <i>Nature Genetics</i> , <b>2014</b> , 46, 61-64	36.3	187
506	Prevalence of vascular complications among patients with glucokinase mutations and prolonged, mild hyperglycemia. <i>JAMA - Journal of the American Medical Association</i> , <b>2014</b> , 311, 279-86	27.4	181
505	The association between postprandial urinary C-peptide creatinine ratio and the treatment response to liraglutide: a multi-centre observational study. <i>Diabetic Medicine</i> , <b>2014</b> , 31, 403-11	3.5	12
504	Permanent neonatal diabetes in siblings with novel C109Y INS mutation transmitted by an unaffected parent with somatic mosaicism. <i>Pediatric Diabetes</i> , <b>2014</b> , 15, 324-8	3.6	4
503	The 0.1% of the population with glucokinase monogenic diabetes can be recognized by clinical characteristics in pregnancy: the Atlantic Diabetes in Pregnancy cohort. <i>Diabetes Care</i> , <b>2014</b> , 37, 1230-6	14.6	90
502	Fetal macrosomia and neonatal hyperinsulinemic hypoglycemia associated with transplacental transfer of sulfonylurea in a mother with KCNJ11-related neonatal diabetes. <i>Diabetes Care</i> , <b>2014</b> , 37, 3333-5	14.6	16
501	Neurogenin 3 is important but not essential for pancreatic islet development in humans. <i>Diabetologia</i> , <b>2014</b> , 57, 2421-4	10.3	24
500	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. <i>Nature Genetics</i> , <b>2014</b> , 46, 812-814	36.3	328
499	A diagnostic approach for defining idiopathic remitting diabetes: a retrospective cohort study. <i>BMC Endocrine Disorders</i> , <b>2014</b> , 14, 45	3.3	
498	Permanent neonatal diabetes misdiagnosed as type 1 diabetes in a 28-year-old female: a life-changing diagnosis. <i>Diabetes Research and Clinical Practice</i> , <b>2014</b> , 106, e22-4	7.4	1
497	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
496	Discovery of biomarkers for glycaemic deterioration before and after the onset of type 2 diabetes: rationale and design of the epidemiological studies within the IMI DIRECT Consortium. <i>Diabetologia</i> , <b>2014</b> , 57, 1132-42	10.3	39
495	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	7 <sup>8</sup> 4

494	Ten years of the national genetic diabetes nurse network: a model for the translation of genetic information into clinical care. <i>Clinical Medicine</i> , <b>2014</b> , 14, 117-21	1.9	9
493	Analysis of transcription factors key for mouse pancreatic development establishes NKX2-2 and MNX1 mutations as causes of neonatal diabetes in man. <i>Cell Metabolism</i> , <b>2014</b> , 19, 146-54	24.6	102
492	Simulation of Finnish population history, guided by empirical genetic data, to assess power of rare-variant tests in Finland. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 710-20	11	19
491	Physiology helps GWAS take a step closer to mechanism. <i>Diabetes</i> , <b>2014</b> , 63, 1836-7	0.9	5
490	Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, 6432-40	5.6	30
489	Effect of the holiday season in patients with diabetes: glycemia and lipids increase postholiday, but the effect is small and transient. <i>Diabetes Care</i> , <b>2014</b> , 37, e98-9	14.6	6
488	Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. <i>PLoS Genetics</i> , <b>2014</b> , 10, e1004123	6	122
487	The HNF4A R76W mutation causes atypical dominant Fanconi syndrome in addition to a Lell phenotype. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 165-9	5.8	57
486	GATA4 mutations are a cause of neonatal and childhood-onset diabetes. <i>Diabetes</i> , <b>2014</b> , 63, 2888-94	0.9	80
485	ISPAD Clinical Practice Consensus Guidelines 2014. The diagnosis and management of monogenic diabetes in children and adolescents. <i>Pediatric Diabetes</i> , <b>2014</b> , 15 Suppl 20, 47-64	3.6	133
484	Home urine C-peptide creatinine ratio can be used to monitor islet transplant function. <i>Diabetes Care</i> , <b>2014</b> , 37, 1737-40	14.6	4
483	Lifecourse: management of type 1 diabetes. <i>Lancet Diabetes and Endocrinology,the</i> , <b>2014</b> , 2, 194-5	18.1	2
482	Cross-sectional and longitudinal studies suggest pharmacological treatment used in patients with glucokinase mutations does not alter glycaemia. <i>Diabetologia</i> , <b>2014</b> , 57, 54-6	10.3	122
481	The majority of patients with long-duration type 1 diabetes are insulin microsecretors and have functioning beta cells. <i>Diabetologia</i> , <b>2014</b> , 57, 187-91	10.3	188
480	Identifying good responders to glucose lowering therapy in type 2 diabetes: implications for stratified medicine. <i>PLoS ONE</i> , <b>2014</b> , 9, e111235	3.7	9
479	Lessons from the mixed-meal tolerance test: use of 90-minute and fasting C-peptide in pediatric diabetes. <i>Diabetes Care</i> , <b>2013</b> , 36, 195-201	14.6	46
478	Improved genetic testing for monogenic diabetes using targeted next-generation sequencing. <i>Diabetologia</i> , <b>2013</b> , 56, 1958-63	10.3	201
477	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. <i>Diabetologia</i> , <b>2013</b> , 56, 758-62	10.3	90

#### (2013-2013)

476	Exome sequencing-driven discovery of coding polymorphisms associated with common metabolic phenotypes. <i>Diabetologia</i> , <b>2013</b> , 56, 298-310	10.3	102
475	Hypoglycaemia following diabetes remission in patients with 6q24 methylation defects: expanding the clinical phenotype. <i>Diabetologia</i> , <b>2013</b> , 56, 218-21	10.3	19
474	Cystatin C is not a good candidate biomarker for HNF1A-MODY. <i>Acta Diabetologica</i> , <b>2013</b> , 50, 815-20	3.9	7
473	The clinical utility of C-peptide measurement in the care of patients with diabetes. <i>Diabetic Medicine</i> , <b>2013</b> , 30, 803-17	3.5	295
472	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. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2013</b> , 98, 4055-62	5.6	219
471	Digenic heterozygous HNF1A and HNF4A mutations in two siblings with childhood-onset diabetes. <i>Pediatric Diabetes</i> , <b>2013</b> , 14, 535-8	3.6	11
470	Continue with long term sulfonylureas in patients with mutations in the KCNJ11 gene when there is evidence of response even if insulin treatment is still required. <i>Diabetes Research and Clinical Practice</i> , <b>2013</b> , 100, e63	7.4	3
469	HNF1B deletions in patients with young-onset diabetes but no known renal disease. <i>Diabetic Medicine</i> , <b>2013</b> , 30, 114-7	3.5	27
468	Home urine C-peptide creatinine ratio (UCPCR) testing can identify type 2 and MODY in pediatric diabetes. <i>Pediatric Diabetes</i> , <b>2013</b> , 14, 181-8	3.6	22
467	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. <i>Nature Genetics</i> , <b>2013</b> , 45, 947-50	36.3	120
466	Five-year follow-up for women with subclinical hypothyroidism in pregnancy. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2013</b> , 98, E1941-5	5.6	34
465	Urine C-peptide creatinine ratio can be used to assess insulin resistance and insulin production in people without diabetes: an observational study. <i>BMJ Open</i> , <b>2013</b> , 3, e003193	3	9
464	Response to Comment on: Chakera et al. Antenatal diagnosis of fetal genotype determines if maternal hyperglycemia due to a glucokinase mutation requires treatment. Diabetes Care 2012;35:1832-1834. <i>Diabetes Care</i> , <b>2013</b> , 36, e15	14.6	2
463	Parental diabetes and birthweight in 236 030 individuals in the UK biobank study. <i>International Journal of Epidemiology</i> , <b>2013</b> , 42, 1714-23	7.8	47
462	A meta-analysis of thyroid-related traits reveals novel loci and gender-specific differences in the regulation of thyroid function. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003266	6	146
461	GATA6 mutations cause a broad phenotypic spectrum of diabetes from pancreatic agenesis to adult-onset diabetes without exocrine insufficiency. <i>Diabetes</i> , <b>2013</b> , 62, 993-7	0.9	104
460	Comment on: Khurana et al. The diagnosis of neonatal diabetes in a mother at 25 years of age. Diabetes Care 2012;35:e59. <i>Diabetes Care</i> , <b>2013</b> , 36, e31	14.6	2
459	Response to comment on: Besser et al. Lessons from the mixed-meal tolerance test: use of 90-minute and fasting C-peptide in pediatric diabetes. Diabetes Care 2013;36:195-201. <i>Diabetes Care</i> , <b>2013</b> , 36, e222	14.6	

458	tRNA methyltransferase homolog gene TRMT10A mutation in young onset diabetes and primary microcephaly in humans. <i>PLoS Genetics</i> , <b>2013</b> , 9, e1003888	6	75
457	Biallelic PDX1 (insulin promoter factor 1) mutations causing neonatal diabetes without exocrine pancreatic insufficiency. <i>Diabetic Medicine</i> , <b>2013</b> , 30, e197-200	3.5	37
456	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
455	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
454	Permanent neonatal diabetes mellitus: prevalence and genetic diagnosis in the SEARCH for Diabetes in Youth Study. <i>Pediatric Diabetes</i> , <b>2013</b> , 14, 174-80	3.6	42
453	Gain-of-function mutations in the K(ATP) channel (KCNJ11) impair coordinated hand-eye tracking. <i>PLoS ONE</i> , <b>2013</b> , 8, e62646	3.7	6
452	Use of HbA1c in the identification of patients with hyperglycaemia caused by a glucokinase mutation: observational case control studies. <i>PLoS ONE</i> , <b>2013</b> , 8, e65326	3.7	83
451	Early-onset, severe lipoatrophy in a patient with permanent neonatal diabetes mellitus secondary to a recessive mutation in the INS gene. <i>Pediatric Diabetes</i> , <b>2012</b> , 13, e26-9	3.6	1
450	KATP channel mutations in infants with permanent diabetes diagnosed after 6 months of life. <i>Pediatric Diabetes</i> , <b>2012</b> , 13, 322-5	3.6	49
449	Incidence, genetics, and clinical phenotype of permanent neonatal diabetes mellitus in northwest Saudi Arabia. <i>Pediatric Diabetes</i> , <b>2012</b> , 13, 499-505	3.6	60
448	Recessive SLC19A2 mutations are a cause of neonatal diabetes mellitus in thiamine-responsive megaloblastic anaemia. <i>Pediatric Diabetes</i> , <b>2012</b> , 13, 314-21	3.6	43
447	Heterozygous ABCC8 mutations are a cause of MODY. <i>Diabetologia</i> , <b>2012</b> , 55, 123-7	10.3	105
446	The impact of insulin administration during the mixed meal tolerance test. <i>Diabetic Medicine</i> , <b>2012</b> , 29, 1279-84	3.5	17
445	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
444	SLC2A2 mutations can cause neonatal diabetes, suggesting GLUT2 may have a role in human insulin secretion. <i>Diabetologia</i> , <b>2012</b> , 55, 2381-5	10.3	82
443	Lipoprotein composition in HNF1A-MODY: differentiating between HNF1A-MODY and type 2 diabetes. <i>Clinica Chimica Acta</i> , <b>2012</b> , 413, 927-32	6.2	30
442	IGF2/H19 hypomethylation in a patient with very low birthweight, preocious pubarche and insulin resistance. <i>BMC Medical Genetics</i> , <b>2012</b> , 13, 42	2.1	22
441	Assessment of endogenous insulin secretion in insulin treated diabetes predicts postprandial glucose and treatment response to prandial insulin. <i>BMC Endocrine Disorders</i> , <b>2012</b> , 12, 6	3.3	10

## (2011-2012)

440	A genome-wide association search for type 2 diabetes genes in African Americans. <i>PLoS ONE</i> , <b>2012</b> , 7, e29202	3.7	138
439	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
438	EDTA improves stability of whole blood C-peptide and insulin to over 24 hours at room temperature. <i>PLoS ONE</i> , <b>2012</b> , 7, e42084	3.7	31
437	The development and validation of a clinical prediction model to determine the probability of MODY in patients with young-onset diabetes. <i>Diabetologia</i> , <b>2012</b> , 55, 1265-72	10.3	172
436	Validation of a single-sample urinary C-peptide creatinine ratio as a reproducible alternative to serum C-peptide in patients with Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2012</b> , 29, 90-3	3.5	22
435	The impact of gender on urine C-peptide creatinine ratio interpretation. <i>Annals of Clinical Biochemistry</i> , <b>2012</b> , 49, 363-8	2.2	10
434	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
433	Novel loci for adiponectin levels and their influence on type 2 diabetes and metabolic traits: a multi-ethnic meta-analysis of 45,891 individuals. <i>PLoS Genetics</i> , <b>2012</b> , 8, e1002607	6	326
432	Amino acid properties may be useful in predicting clinical outcome in patients with Kir6.2 neonatal diabetes. <i>European Journal of Endocrinology</i> , <b>2012</b> , 167, 417-21	6.5	3
431	Identifying clinical criteria to predict Type 1 diabetes, as defined by absolute insulin deficiency: a systematic review protocol. <i>BMJ Open</i> , <b>2012</b> , 2,	3	3
430	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
429	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
428	Permanent neonatal diabetes: different aetiology in Arabs compared to Europeans. <i>Archives of Disease in Childhood</i> , <b>2012</b> , 97, 721-3	2.2	25
427	Antenatal diagnosis of fetal genotype determines if maternal hyperglycemia due to a glucokinase mutation requires treatment. <i>Diabetes Care</i> , <b>2012</b> , 35, 1832-4	14.6	41
426	Genetic variation in the 15q25 nicotinic acetylcholine receptor gene cluster (CHRNA5-CHRNA3-CHRNB4) interacts with maternal self-reported smoking status during pregnancy to influence birth weight. <i>Human Molecular Genetics</i> , <b>2012</b> , 21, 5344-58	5.6	50
425	The role of inflammatory pathway genetic variation on maternal metabolic phenotypes during pregnancy. <i>PLoS ONE</i> , <b>2012</b> , 7, e32958	3.7	19
424	Using highly sensitive C-reactive protein measurement to diagnose MODY in a family with suspected type 2 diabetes. <i>BMJ Case Reports</i> , <b>2012</b> , 2012,	0.9	
423	GATA6 haploinsufficiency causes pancreatic agenesis in humans. <i>Nature Genetics</i> , <b>2011</b> , 44, 20-22	36.3	195

422	Urinary C-peptide creatinine ratio is a practical outpatient tool for identifying hepatocyte nuclear factor 1-{alpha}/hepatocyte nuclear factor 4-{alpha} maturity-onset diabetes of the young from long-duration type 1 diabetes. <i>Diabetes Care</i> , <b>2011</b> , 34, 286-91	14.6	96
421	Permanent Neonatal Diabetes and Enteric Anendocrinosis Associated With Biallelic Mutations in NEUROG3. <i>Diabetes</i> , <b>2011</b> , 60, 1349-53	0.9	114
420	A pathway to insulin independence in newborns and infants with diabetes. <i>Journal of Perinatology</i> , <b>2011</b> , 31, 567-70	3.1	1
419	High-sensitivity CRP discriminates HNF1A-MODY from other subtypes of diabetes. <i>Diabetes Care</i> , <b>2011</b> , 34, 1860-2	14.6	76
418	Response to Comment on: McDonald et al. High-Sensitivity CRP Discriminates HNF1A-MODY From Other Subtypes of Diabetes. Diabetes Care 2011;34:1860-1862. <i>Diabetes Care</i> , <b>2011</b> , 34, e187-e187	14.6	78
417	Persistently autoantibody negative (PAN) type 1 diabetes mellitus in children. <i>Pediatric Diabetes</i> , <b>2011</b> , 12, 142-9	3.6	44
416	KCNJ11 activating mutations cause both transient and permanent neonatal diabetes mellitus in Cypriot patients. <i>Pediatric Diabetes</i> , <b>2011</b> , 12, 133-7	3.6	5
415	Sequencing PDX1 (insulin promoter factor 1) in 1788 UK individuals found 5% had a low frequency coding variant, but these variants are not associated with Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2011</b> , 28, 681-4	3.5	10
414	Urine C-peptide creatinine ratio is an alternative to stimulated serum C-peptide measurement in late-onset, insulin-treated diabetes. <i>Diabetic Medicine</i> , <b>2011</b> , 28, 1034-8	3.5	26
413	Islet autoantibodies can discriminate maturity-onset diabetes of the young (MODY) from Type 1 diabetes. <i>Diabetic Medicine</i> , <b>2011</b> , 28, 1028-33	3.5	127
412	Genome-wide association scan allowing for epistasis in type 2 diabetes. <i>Annals of Human Genetics</i> , <b>2011</b> , 75, 10-9	2.2	29
411	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
410	A conserved tryptophan at the membrane-water interface acts as a gatekeeper for Kir6.2/SUR1 channels and causes neonatal diabetes when mutated. <i>Journal of Physiology</i> , <b>2011</b> , 589, 3071-83	3.9	13
409	Common nonsynonymous substitutions in SLCO1B1 predispose to statin intolerance in routinely treated individuals with type 2 diabetes: a go-DARTS study. <i>Clinical Pharmacology and Therapeutics</i> , <b>2011</b> , 89, 210-6	6.1	153
408	A role for coding functional variants in HNF4A in type 2 diabetes susceptibility. <i>Diabetologia</i> , <b>2011</b> , 54, 111-9	10.3	24
407	The previously reported T342P GCK missense variant is not a pathogenic mutation causing MODY. <i>Diabetologia</i> , <b>2011</b> , 54, 2202-5	10.3	11
406	Fetal thyroid hormone level at birth is associated with fetal growth. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2011</b> , 96, E934-8	5.6	75
405	Novel GLIS3 mutations demonstrate an extended multisystem phenotype. <i>European Journal of Endocrinology</i> , <b>2011</b> , 164, 437-43	6.5	75

## (2010-2011)

404	A meta-analysis of the associations between common variation in the PDE8B gene and thyroid hormone parameters, including assessment of longitudinal stability of associations over time and effect of thyroid hormone replacement. <i>European Journal of Endocrinology</i> , <b>2011</b> , 164, 773-80	6.5	26
403	Genetic variation at CHRNA5-CHRNA3-CHRNB4 interacts with smoking status to influence body mass index. <i>International Journal of Epidemiology</i> , <b>2011</b> , 40, 1617-28	7.8	92
402	Mendelian randomization studies do not support a role for raised circulating triglyceride levels influencing type 2 diabetes, glucose levels, or insulin resistance. <i>Diabetes</i> , <b>2011</b> , 60, 1008-18	0.9	60
401	Urine C-peptide creatinine ratio is a noninvasive alternative to the mixed-meal tolerance test in children and adults with type 1 diabetes. <i>Diabetes Care</i> , <b>2011</b> , 34, 607-9	14.6	48
400	Mutations of the same conserved glutamate residue in NBD2 of the sulfonylurea receptor 1 subunit of the KATP channel can result in either hyperinsulinism or neonatal diabetes. <i>Diabetes</i> , <b>2011</b> , 60, 1813-22	0.9	20
399	Loss-of-function CYP2C9 variants improve therapeutic response to sulfonylureas in type 2 diabetes: a Go-DARTS study. <i>Clinical Pharmacology and Therapeutics</i> , <b>2010</b> , 87, 52-6	6.1	124
398	Rfx6 directs islet formation and insulin production in mice and humans. <i>Nature</i> , <b>2010</b> , 463, 775-80	50.4	254
397	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
396	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
395	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
394	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
393	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
392	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
391	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
390	Increased all-cause and cardiovascular mortality in monogenic diabetes as a result of mutations in the HNF1A gene. <i>Diabetic Medicine</i> , <b>2010</b> , 27, 157-61	3.5	76
389	Incorrect and incomplete coding and classification of diabetes: a systematic review. <i>Diabetic Medicine</i> , <b>2010</b> , 27, 491-7	3.5	47
388	Entities and frequency of neonatal diabetes: data from the diabetes documentation and quality management system (DPV). <i>Diabetic Medicine</i> , <b>2010</b> , 27, 709-12	3.5	63
387	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. <i>Pediatric Diabetes</i> , <b>2010</b> , 11, 18-23	3.6	42

386	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
385	Genetic Testing in Diabetes Mellitus <b>2010</b> , 17-25		3
384	Interaction between mutations in the slide helix of Kir6.2 associated with neonatal diabetes and neurological symptoms. <i>Human Molecular Genetics</i> , <b>2010</b> , 19, 963-72	5.6	15
383	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2010</b> , 107, 3105-10	11.5	149
382	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
381	Hyperglycemia and Adverse Pregnancy Outcome (HAPO) study: common genetic variants in GCK and TCF7L2 are associated with fasting and postchallenge glucose levels in pregnancy and with the new consensus definition of gestational diabetes mellitus from the International Association of	0.9	77
380	Detailed investigation of the role of common and low-frequency WFS1 variants in type 2 diabetes risk. <i>Diabetes</i> , <b>2010</b> , 59, 741-6	0.9	27
379	Genetic influences on the association between fetal growth and susceptibility to type 2 diabetes. Journal of Developmental Origins of Health and Disease, <b>2010</b> , 1, 96-105	2.4	6
378	Polygenic risk variants for type 2 diabetes susceptibility modify age at diagnosis in monogenic HNF1A diabetes. <i>Diabetes</i> , <b>2010</b> , 59, 266-71	0.9	25
377	Homozygous mutations in NEUROD1 are responsible for a novel syndrome of permanent neonatal diabetes and neurological abnormalities. <i>Diabetes</i> , <b>2010</b> , 59, 2326-31	0.9	116
376	Reevaluation of a case of type 1 diabetes mellitus diagnosed before 6 months of age. <i>Nature Reviews Endocrinology</i> , <b>2010</b> , 6, 347-51	15.2	6
375	New genetic loci implicated in fasting glucose homeostasis and their impact on type 2 diabetes risk. <i>Nature Genetics</i> , <b>2010</b> , 42, 105-16	36.3	1673
374	Genetic association analysis of LARS2 with type 2 diabetes. <i>Diabetologia</i> , <b>2010</b> , 53, 103-10	10.3	8
373	Maturity-onset diabetes of the young (MODY): how many cases are we missing?. <i>Diabetologia</i> , <b>2010</b> , 53, 2504-8	10.3	432
372	Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. <i>Human Genetics</i> , <b>2010</b> , 127, 55-64	6.3	44
371	KCNJ11 activating mutation in an Indian family with remitting and relapsing diabetes. <i>Indian Journal of Pediatrics</i> , <b>2010</b> , 77, 551-4	3	6
370	Mutations in the hepatocyte nuclear factor-1[(HNF1B) gene are common with combined uterine and renal malformations but are not found with isolated uterine malformations. <i>American Journal of Obstetrics and Gynecology</i> , <b>2010</b> , 203, 364.e1-5	6.4	39
369	A powerful approach to sub-phenotype analysis in population-based genetic association studies. <i>Genetic Epidemiology</i> , <b>2010</b> , 34, 335-43	2.6	39

## (2009-2010)

368	Sequencing of candidate genes selected by beta cell experts in monogenic diabetes of unknown aetiology. <i>JOP: Journal of the Pancreas</i> , <b>2010</b> , 11, 14-7	1.2	8
367	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
366	Dietary energy density affects fat mass in early adolescence and is not modified by FTO variants. <i>PLoS ONE</i> , <b>2009</b> , 4, e4594	3.7	50
365	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
364	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
363	Underlying genetic models of inheritance in established type 2 diabetes associations. <i>American Journal of Epidemiology</i> , <b>2009</b> , 170, 537-45	3.8	60
362	Stability and reproducibility of a single-sample urinary C-peptide/creatinine ratio and its correlation with 24-h urinary C-peptide. <i>Clinical Chemistry</i> , <b>2009</b> , 55, 2035-9	5.5	48
361	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	0.9	98
360	Linkage disequilibrium mapping of the replicated type 2 diabetes linkage signal on chromosome 1q. <i>Diabetes</i> , <b>2009</b> , 58, 1704-9	0.9	23
359	Phosphodiesterase 8B gene polymorphism is associated with subclinical hypothyroidism in pregnancy. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2009</b> , 94, 4608-12	5.6	27
358	Tooth discoloration in patients with neonatal diabetes after transfer onto glibenclamide: a previously unreported side effect. <i>Diabetes Care</i> , <b>2009</b> , 32, 1428-30	14.6	35
357	RET gene mutations are not a common cause of congenital solitary functioning kidney in adults. <i>CKJ: Clinical Kidney Journal</i> , <b>2009</b> , 2, 183-4	4.5	
356	Wolcott-Rallison syndrome is the most common genetic cause of permanent neonatal diabetes in consanguineous families. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2009</b> , 94, 4162-70	5.6	103
355	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. <i>Human Molecular Genetics</i> , <b>2009</b> , 18, 2922-7	5.6	122
354	Type 2 diabetes risk alleles are associated with reduced size at birth. <i>Diabetes</i> , <b>2009</b> , 58, 1428-33	0.9	117
353	Common variation in the DIO2 gene predicts baseline psychological well-being and response to combination thyroxine plus triiodothyronine therapy in hypothyroid patients. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2009</b> , 94, 1623-9	5.6	224
352	Cigarette smoking during pregnancy is associated with alterations in maternal and fetal thyroid function. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2009</b> , 94, 570-4	5.6	51
351	Adjacent mutations in the gating loop of Kir6.2 produce neonatal diabetes and hyperinsulinism. <i>EMBO Molecular Medicine</i> , <b>2009</b> , 1, 166-77	12	34

350	The association of common genetic variants in the APOA5, LPL and GCK genes with longitudinal changes in metabolic and cardiovascular traits. <i>Diabetologia</i> , <b>2009</b> , 52, 106-14	10.3	22
349	Glucokinase, the pancreatic glucose sensor, is not the gut glucose sensor. <i>Diabetologia</i> , <b>2009</b> , 52, 154-9	10.3	33
348	FTO gene variants are strongly associated with type 2 diabetes in South Asian Indians. <i>Diabetologia</i> , <b>2009</b> , 52, 247-52	10.3	145
347	Referral rates for diagnostic testing support an incidence of permanent neonatal diabetes in three European countries of at least 1 in 260,000 live births. <i>Diabetologia</i> , <b>2009</b> , 52, 1683-5	10.3	98
346	Circulating beta-carotene levels and type 2 diabetes-cause or effect?. <i>Diabetologia</i> , <b>2009</b> , 52, 2117-21	10.3	21
345	FTO gene variation and measures of body mass in an African population. <i>BMC Medical Genetics</i> , <b>2009</b> , 10, 21	2.1	77
344	Mutations in the ABCC8 (SUR1 subunit of the K(ATP) channel) gene are associated with a variable clinical phenotype. <i>Clinical Endocrinology</i> , <b>2009</b> , 71, 358-62	3.4	30
343	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
342	Definition, epidemiology and classification of diabetes in children and adolescents. <i>Pediatric Diabetes</i> , <b>2009</b> , 10 Suppl 12, 3-12	3.6	169
341	The diagnosis and management of monogenic diabetes in children and adolescents. <i>Pediatric Diabetes</i> , <b>2009</b> , 10 Suppl 12, 33-42	3.6	205
340	Mutations in the third gene shown to alter fasting glucose levels in the population (G6PC2) are not a common cause of monogenic forms of pancreatic B-cell dysfunction. <i>Diabetic Medicine</i> , <b>2009</b> , 26, 113-	43.5	2
339	Pregnancy outcome in patients with raised blood glucose due to a heterozygous glucokinase gene mutation. <i>Diabetic Medicine</i> , <b>2009</b> , 26, 14-8	3.5	94
338	A genetic diagnosis of HNF1A diabetes alters treatment and improves glycaemic control in the majority of insulin-treated patients. <i>Diabetic Medicine</i> , <b>2009</b> , 26, 437-41	3.5	168
337	Testing for monogenic diabetes among children and adolescents with antibody-negative clinically defined Type 1 diabetes. <i>Diabetic Medicine</i> , <b>2009</b> , 26, 1070-4	3.5	44
336	Neonatal hyperinsulinaemic hypoglycaemia and monogenic diabetes due to a heterozygous mutation of the HNF4A gene. <i>Australian and New Zealand Journal of Obstetrics and Gynaecology</i> , <b>2009</b> , 49, 328-30	1.7	13
335	Clinical heterogeneity in patients with FOXP3 mutations presenting with permanent neonatal diabetes. <i>Diabetes Care</i> , <b>2009</b> , 32, 111-6	14.6	86
334	Hypoplastic glomerulocystic kidney disease and hepatoblastoma: a potential association not caused by mutations in hepatocyte nuclear factor 1beta. <i>Journal of Pediatric Hematology/Oncology</i> , <b>2009</b> , 31, 527-9	1.2	7
333	Low frequency variants in the exons only encoding isoform A of HNF1A do not contribute to susceptibility to type 2 diabetes. <i>PLoS ONE</i> , <b>2009</b> , 4, e6615	3.7	4

## (2008-2008)

332	A mutation (R826W) in nucleotide-binding domain 1 of ABCC8 reduces ATPase activity and causes transient neonatal diabetes. <i>EMBO Reports</i> , <b>2008</b> , 9, 648-54	6.5	35
331	Genome-wide association analysis identifies 20 loci that influence adult height. <i>Nature Genetics</i> , <b>2008</b> , 40, 575-83	36.3	654
330	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
329	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. <i>Nature Genetics</i> , <b>2008</b> , 40, 949-51	36.3	417
328	Sulphonylurea therapy improves cognition in a patient with the V59M KCNJ11 mutation. <i>Diabetic Medicine</i> , <b>2008</b> , 25, 277-81	3.5	89
327	Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. <i>Diabetic Medicine</i> , <b>2008</b> , 25, 383-99	3.5	178
326	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
325	Learning from molecular genetics: novel insights arising from the definition of genes for monogenic and type 2 diabetes. <i>Diabetes</i> , <b>2008</b> , 57, 2889-98	0.9	96
324	Clinical implications of a molecular genetic classification of monogenic beta-cell diabetes. <i>Nature Clinical Practice Endocrinology and Metabolism</i> , <b>2008</b> , 4, 200-13		367
323	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
322	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
321	Mutations in the glucokinase gene of the fetus result in reduced placental weight. <i>Diabetes Care</i> , <b>2008</b> , 31, 753-7	14.6	20
320	Effective treatment with oral sulfonylureas in patients with diabetes due to sulfonylurea receptor 1 (SUR1) mutations. <i>Diabetes Care</i> , <b>2008</b> , 31, 204-9	14.6	203
319	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
318	Population-specific risk of type 2 diabetes conferred by HNF4A P2 promoter variants: a lesson for replication studies. <i>Diabetes</i> , <b>2008</b> , 57, 3161-5	0.9	33
317	Persistent hyperinsulinemic hypoglycemia and maturity-onset diabetes of the young due to heterozygous HNF4A mutations. <i>Diabetes</i> , <b>2008</b> , 57, 1659-63	0.9	113
316	Coincidence of a novel KCNJ11 missense variant R365H with a paternally inherited 6q24 duplication in a patient with transient neonatal diabetes. <i>Diabetes Care</i> , <b>2008</b> , 31, 1736-7	14.6	1
315	PREVALENCE AND INCIDENCE OF A NEWLY DEFINED TYPE OF DIABETES IN CHILDREN, ADOLESCENTS, AND ADULTS IN THE LARGEST INTERNATIONAL SERIES TO DATE. <i>Pediatrics</i> , <b>2008</b> , 121, S115.3-S116	7.4	

314	The diabetic phenotype in HNF4A mutation carriers is moderated by the expression of HNF4A isoforms from the P1 promoter during fetal development. <i>Diabetes</i> , <b>2008</b> , 57, 1745-52	0.9	55
313	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
312	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
311	Diabetes susceptibility in the Canadian Oji-Cree population is moderated by abnormal mRNA processing of HNF1A G319S transcripts. <i>Diabetes</i> , <b>2008</b> , 57, 1978-82	0.9	23
310	A common variation in deiodinase 1 gene DIO1 is associated with the relative levels of free thyroxine and triiodothyronine. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2008</b> , 93, 3075-81	5.6	102
309	Severe intrauterine growth retardation and atypical diabetes associated with a translocation breakpoint disrupting regulation of the insulin-like growth factor 2 gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2008</b> , 93, 4373-80	5.6	30
308	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. <i>Diabetes</i> , <b>2008</b> , 57, 1034-42	0.9	299
307	A genetic syndrome of chronic renal failure with multiple renal cysts and early onset diabetes. <i>Kidney International</i> , <b>2008</b> , 74, 1094-9	9.9	14
306	The fat mass- and obesity-associated locus and dietary intake in children. <i>American Journal of Clinical Nutrition</i> , <b>2008</b> , 88, 971-8	7	213
305	Glibenclamide unresponsiveness in a Brazilian child with permanent neonatal diabetes mellitus and DEND syndrome due to a C166Y mutation in KCNJ11 (Kir6.2) gene. <i>Arquivos Brasileiros De Endocrinologia E Metabologia</i> , <b>2008</b> , 52, 1350-5		16
304	A Kir6.2 mutation causing severe functional effects in vitro produces neonatal diabetes without the expected neurological complications. <i>Diabetologia</i> , <b>2008</b> , 51, 802-10	10.3	25
303	Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. <i>Diabetologia</i> , <b>2008</b> , 51, 546-53	10.3	311
302	Gene variants influencing measures of inflammation or predisposing to autoimmune and inflammatory diseases are not associated with the risk of type 2 diabetes. <i>Diabetologia</i> , <b>2008</b> , 51, 2205-	130.3	38
301	Hepatocyte nuclear factor-1beta gene deletionsa common cause of renal disease. <i>Nephrology Dialysis Transplantation</i> , <b>2008</b> , 23, 627-35	4.3	79
300	The genetics of diabetic pregnancy. Series in Maternal-fetal Medicine, 2008, 466-474		
299	Genetic Disorders of the Pancreatic Beta Cell and Diabetes (Permanent Neonatal Diabetes and Maturity-Onset Diabetes of the Young) <b>2008</b> , 399-430		6
298	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
297	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	5.6	7

## (2007-2007)

296	Macrosomia and hyperinsulinaemic hypoglycaemia in patients with heterozygous mutations in the HNF4A gene. <i>PLoS Medicine</i> , <b>2007</b> , 4, e118	11.6	279
295	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
294	Common variants in WFS1 confer risk of type 2 diabetes. <i>Nature Genetics</i> , <b>2007</b> , 39, 951-3	36.3	296
293	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
292	Genetics and type 2 diabetes in youth. <i>Pediatric Diabetes</i> , <b>2007</b> , 8 Suppl 9, 42-7	3.6	17
291	Mutations in the ABCC8 gene encoding the SUR1 subunit of the KATP channel cause transient neonatal diabetes, permanent neonatal diabetes or permanent diabetes diagnosed outside the neonatal period. <i>Diabetes, Obesity and Metabolism</i> , <b>2007</b> , 9 Suppl 2, 28-39	6.7	67
290	Low prevalence of mitochondrial DNA 3243A>G point mutation in Caucasians with unexplained renal disease. <i>Diabetic Medicine</i> , <b>2007</b> , 24, 804-6	3.5	
289	Reduced peripheral blood mitochondrial DNA content is not a risk factor for Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2007</b> , 24, 784-7	3.5	15
288	The transcription factor 7-like 2 (TCF7L2) gene is associated with Type 2 diabetes in UK community-based cases, but the risk allele frequency is reduced compared with UK cases selected for genetic studies. <i>Diabetic Medicine</i> , <b>2007</b> , 24, 1067-72	3.5	15
287	A common variant of the p16(INK4a) genetic region is associated with physical function in older people. <i>Mechanisms of Ageing and Development</i> , <b>2007</b> , 128, 370-7	5.6	75
286	The long-term impact on offspring of exposure to hyperglycaemia in utero due to maternal glucokinase gene mutations. <i>Diabetologia</i> , <b>2007</b> , 50, 620-4	10.3	30
285	TCF7L2 in the Go-DARTS study: evidence for a gene dose effect on both diabetes susceptibility and control of glucose levels. <i>Diabetologia</i> , <b>2007</b> , 50, 1186-91	10.3	65
284	Mutations in HHEX are not a common cause of monogenic forms of beta cell dysfunction. <i>Diabetologia</i> , <b>2007</b> , 50, 2019-2022	10.3	3
283	Partial and whole gene deletion mutations of the GCK and HNF1A genes in maturity-onset diabetes of the young. <i>Diabetologia</i> , <b>2007</b> , 50, 2313-7	10.3	54
282	Common variants in the TCF7L2 gene are strongly associated with type 2 diabetes mellitus in the Indian population. <i>Diabetologia</i> , <b>2007</b> , 50, 63-7	10.3	191
281	An association analysis of the HLA gene region in latent autoimmune diabetes in adults. <i>Diabetologia</i> , <b>2007</b> , 50, 68-73	10.3	62
280	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. <i>Diabetes</i> , <b>2007</b> , 56, 1930-7	0.9	273
279	Variation in TCF7L2 influences therapeutic response to sulfonylureas: a GoDARTs study. <i>Diabetes</i> , <b>2007</b> , 56, 2178-82	0.9	251

278	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
277	Origin of de novo KCNJ11 mutations and risk of neonatal diabetes for subsequent siblings. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2007</b> , 92, 1773-7	5.6	46
276	Increased ATPase activity produced by mutations at arginine-1380 in nucleotide-binding domain 2 of ABCC8 causes neonatal diabetes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 18988-92	11.5	47
275	The impact of maternal glycemia and obesity on early postnatal growth in a nondiabetic Caucasian population. <i>Diabetes Care</i> , <b>2007</b> , 30, 777-83	14.6	53
274	Measurement of cord insulin and insulin-related peptides suggests that girls are more insulin resistant than boys at birth. <i>Diabetes Care</i> , <b>2007</b> , 30, 2661-6	14.6	59
273	Prevalence of permanent neonatal diabetes in Slovakia and successful replacement of insulin with sulfonylurea therapy in KCNJ11 and ABCC8 mutation carriers. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2007</b> , 92, 1276-82	5.6	82
272	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
271	Insulin gene mutations as a cause of permanent neonatal diabetes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2007</b> , 104, 15040-4	11.5	426
270	A novel mutation causing DEND syndrome: a treatable channelopathy of pancreas and brain. <i>Neurology</i> , <b>2007</b> , 69, 1342-9	6.5	91
269	Sulfonylurea treatment in young children with neonatal diabetes: dealing with hyperglycemia, hypoglycemia, and sick days. <i>Diabetes Care</i> , <b>2007</b> , 30, e28-9	14.6	28
268	Mitochondrial DNA haplogroups and type 2 diabetes: a study of 897 cases and 1010 controls. Journal of Medical Genetics, <b>2007</b> , 44, e80	5.8	45
267	Common variation in the LMNA gene (encoding lamin A/C) and type 2 diabetes: association analyses in 9,518 subjects. <i>Diabetes</i> , <b>2007</b> , 56, 879-83	0.9	27
266	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
265	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. <i>American Journal of Human Genetics</i> , <b>2007</b> , 81, 375-82	11	161
264	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
263	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
262	Prime suspect: the TCF7L2 gene and type 2 diabetes risk. <i>Journal of Clinical Investigation</i> , <b>2007</b> , 117, 207	′ <b>75</b> 99	37
261	Common variations in the ALMS1 gene do not contribute to susceptibility to type 2 diabetes in a large white UK population. <i>Diabetologia</i> , <b>2006</b> , 49, 1209-13	10.3	17

#### (2006-2006)

260	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. <i>Diabetologia</i> , <b>2006</b> , 49, 1190-7	10.3	190
259	Paternal insulin resistance and its association with umbilical cord insulin concentrations. <i>Diabetologia</i> , <b>2006</b> , 49, 2668-74	10.3	16
258	Reduced beta cell function in offspring of mothers with young-onset type 2 diabetes. <i>Diabetologia</i> , <b>2006</b> , 49, 1876-80	10.3	22
257	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.	10.3	105
256	Offspring birthweight is not associated with paternal insulin resistance. <i>Diabetologia</i> , <b>2006</b> , 49, 2675-8	10.3	12
255	Determinants of insulin concentrations in healthy 1-week-old babies in the community: applications of a bloodspot assay. <i>Early Human Development</i> , <b>2006</b> , 82, 143-8	2.2	10
254	Assessing newborn body composition using principal components analysis: differences in the determinants of fat and skeletal size. <i>BMC Pediatrics</i> , <b>2006</b> , 6, 24	2.6	20
253	Contrasting insulin sensitivity of endogenous glucose production rate in subjects with hepatocyte nuclear factor-1beta and -1alpha mutations. <i>Diabetes</i> , <b>2006</b> , 55, 405-11	0.9	39
252	Combining information from common type 2 diabetes risk polymorphisms improves disease prediction. <i>PLoS Medicine</i> , <b>2006</b> , 3, e374	11.6	214
251	Conditional expression of hepatocyte nuclear factor-1beta, the maturity-onset diabetes of the young-5 gene product, influences the viability and functional competence of pancreatic beta-cells. Journal of Endocrinology, <b>2006</b> , 190, 171-81	4.7	18
250	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 1793-800	5.6	175
249	Significant linkage of BMI to chromosome 10p in the U.K. population and evaluation of GAD2 as a positional candidate. <i>Diabetes</i> , <b>2006</b> , 55, 1884-9	0.9	22
248	Variation within the gene encoding the upstream stimulatory factor 1 does not influence susceptibility to type 2 diabetes in samples from populations with replicated evidence of linkage to chromosome 1q. <i>Diabetes</i> , <b>2006</b> , 55, 2541-8	0.9	33
247	Assessment of the role of common genetic variation in the transient neonatal diabetes mellitus (TNDM) region in type 2 diabetes: a comparative genomic and tagging single nucleotide polymorphism approach. <i>Diabetes</i> , <b>2006</b> , 55, 2272-6	0.9	10
246	The variable number of tandem repeats upstream of the insulin gene is a susceptibility locus for latent autoimmune diabetes in adults. <i>Diabetes</i> , <b>2006</b> , 55, 1890-4	0.9	40
245	Minireview: pharmacogenetics and beyond: the interaction of therapeutic response, beta-cell physiology, and genetics in diabetes. <i>Endocrinology</i> , <b>2006</b> , 147, 2657-63	4.8	101
244	No evidence of association of ENPP1 variants with type 2 diabetes or obesity in a study of 8,089 U.K. Caucasians. <i>Diabetes</i> , <b>2006</b> , 55, 3175-9	0.9	75
243	Phenotypic multiple endocrine neoplasia type 2B, without endocrinopathy or RET gene mutation: implications for management. <i>Thyroid</i> , <b>2006</b> , 16, 605-8	6.2	8

242	Mutations at the same residue (R50) of Kir6.2 (KCNJ11) that cause neonatal diabetes produce different functional effects. <i>Diabetes</i> , <b>2006</b> , 55, 1705-12	0.9	56
241	A Kir6.2 mutation causing neonatal diabetes impairs electrical activity and insulin secretion from INS-1 beta-cells. <i>Diabetes</i> , <b>2006</b> , 55, 3075-82	0.9	37
240	Activating mutations in the gene encoding Kir6.2 alter fetal and postnatal growth and also cause neonatal diabetes. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2006</b> , 91, 2782-8	5.6	32
239	HLA genotyping supports a nonautoimmune etiology in patients diagnosed with diabetes under the age of 6 months. <i>Diabetes</i> , <b>2006</b> , 55, 1895-8	0.9	92
238	Chapter 1 Transcription factor genes in type 2 diabetes. <i>Advances in Molecular and Cellular Endocrinology</i> , <b>2006</b> , 5, 1-14		2
237	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
236	Mutations in hepatocyte nuclear factor-1beta and their related phenotypes. <i>Journal of Medical Genetics</i> , <b>2006</b> , 43, 84-90	5.8	236
235	Allelic drop-out may occur with a primer binding site polymorphism for the commonly used RFLP assay for the -1131T>C polymorphism of the Apolipoprotein AV gene. <i>Lipids in Health and Disease</i> , <b>2006</b> , 5, 11	4.4	11
234	Rapid and sensitive real-time polymerase chain reaction method for detection and quantification of 3243A>G mitochondrial point mutation. <i>Journal of Molecular Diagnostics</i> , <b>2006</b> , 8, 225-30	5.1	22
233	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. <i>Human Molecular Genetics</i> , <b>2006</b> , 15, 2216-24	5.6	100
232	A common haplotype of the glucokinase gene alters fasting glucose and birth weight: association in six studies and population-genetics analyses. <i>American Journal of Human Genetics</i> , <b>2006</b> , 79, 991-1001	11	103
231	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. <i>New England Journal of Medicine</i> , <b>2006</b> , 355, 467-77	59.2	740
230	Association of the calpain-10 gene with type 2 diabetes in Europeans: results of pooled and meta-analyses. <i>Molecular Genetics and Metabolism</i> , <b>2006</b> , 89, 174-84	3.7	69
229	The functional "KL-VS" variant of KLOTHO is not associated with type 2 diabetes in 5028 UK Caucasians. <i>BMC Medical Genetics</i> , <b>2006</b> , 7, 51	2.1	15
228	A study of association between common variation in the growth hormone-chorionic somatomammotropin hormone gene cluster and adult fasting insulin in a UK Caucasian population. <i>Journal of Negative Results in BioMedicine</i> , <b>2006</b> , 5, 18		1
227	ISPAD Clinical Practice Consensus Guidelines 2006-2007. Definition, epidemiology and classification. <i>Pediatric Diabetes</i> , <b>2006</b> , 7, 343-51	3.6	57
226	ISPAD Clinical Practice Consensus Guidelines 2006-2007. The diagnosis and management of monogenic diabetes in children. <i>Pediatric Diabetes</i> , <b>2006</b> , 7, 352-60	3.6	108
225	The effect of obesity on glycaemic response to metformin or sulphonylureas in Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 128-33	3.5	63

224	Familial factors in diabetic nephropathy: an offspring study. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 331-4	3.5	26
223	GAD antibodies in probands and their relatives in a cohort clinically selected for Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 834-8	3.5	16
222	Asian MODY: are we missing an important diagnosis?. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 1257-60	3.5	17
221	Maternal glucose levels influence birthweight and Natch-upNand Natch-downNgrowth in a large contemporary cohort. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 1207-12	3.5	11
220	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. <i>Diabetic Medicine</i> , <b>2006</b> , 23, 1301-6	3.5	123
219	Epistasis between type 2 diabetes susceptibility Loci on chromosomes 1q21-25 and 10q23-26 in northern Europeans. <i>Annals of Human Genetics</i> , <b>2006</b> , 70, 726-37	2.2	16
218	The Exeter Family Study of Childhood Health (EFSOCH): study protocol and methodology. <i>Paediatric and Perinatal Epidemiology</i> , <b>2006</b> , 20, 172-9	2.7	46
217	KCNJ11 activating mutations are associated with developmental delay, epilepsy and neonatal diabetes syndrome and other neurological features. <i>European Journal of Human Genetics</i> , <b>2006</b> , 14, 824	I- <b>3</b> 6	110
216	The impact of the angiotensin-converting enzyme insertion/deletion polymorphism on severe hypoglycemia in Type 2 diabetes. <i>Review of Diabetic Studies</i> , <b>2006</b> , 3, 76-81	3.6	9
215	Role of the mitochondrial DNA 16184-16193 poly-C tract in type 2 diabetes. <i>Lancet, The</i> , <b>2005</b> , 366, 165	50 <sub>4</sub> 15	59
214	What makes a good genetic association study?. Lancet, The, 2005, 366, 1315-23	40	408
213	C-reactive protein and its role in metabolic syndrome: mendelian randomisation study. <i>Lancet, The</i> , <b>2005</b> , 366, 1954-9	40	266
212	Mutations in the Kir6.2 subunit of the KATP channel and permanent neonatal diabetes: new insights and new treatment. <i>Annals of Medicine</i> , <b>2005</b> , 37, 186-95	1.5	75
211	Permanent neonatal diabetes in an Asian infant. <i>Journal of Pediatrics</i> , <b>2005</b> , 146, 131-3	3.6	48
210	Reported parental age of death in type 2 diabetic patients with and without established diabetic nephropathy. <i>European Journal of Internal Medicine</i> , <b>2005</b> , 16, 482-4	3.9	
209	Preserved insulin response to tolbutamide in hepatocyte nuclear factor-1alpha mutation carriers. <i>Diabetic Medicine</i> , <b>2005</b> , 22, 406-9	3.5	12
208	Examining the relationships between the Pro12Ala variant in PPARG and Type 2 diabetes-related traits in UK samples. <i>Diabetic Medicine</i> , <b>2005</b> , 22, 1696-700	3.5	19
207	Detection of an MEN1 gene mutation depends on clinical features and supports current referral criteria for diagnostic molecular genetic testing. <i>Clinical Endocrinology</i> , <b>2005</b> , 62, 169-75	3.4	79

206	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
205	A gating mutation at the internal mouth of the Kir6.2 pore is associated with DEND syndrome. <i>EMBO Reports</i> , <b>2005</b> , 6, 470-5	6.5	95
204	Evidence of genetic regulation of fetal longitudinal growth. <i>Early Human Development</i> , <b>2005</b> , 81, 823-31	l 2.2	63
203	KCNJ11 activating mutations in Italian patients with permanent neonatal diabetes. <i>Human Mutation</i> , <b>2005</b> , 25, 22-7	4.7	118
202	The identification of a R201H mutation in KCNJ11, which encodes Kir6.2, and successful transfer to sustained-release sulphonylurea therapy in a subject with neonatal diabetes: evidence for heterogeneity of beta cell function among carriers of the R201H mutation. <i>Diabetologia</i> , <b>2005</b> , 48, 1029	10.3 <b>)-31</b>	69
201	Molecular genetics and phenotypic characteristics of MODY caused by hepatocyte nuclear factor 4alpha mutations in a large European collection. <i>Diabetologia</i> , <b>2005</b> , 48, 878-85	10.3	170
200	Large-scale studies of the association between variation at the TNF/LTA locus and susceptibility to type 2 diabetes. <i>Diabetologia</i> , <b>2005</b> , 48, 2013-7	10.3	40
199	The position of premature termination codons in the hepatocyte nuclear factor -1 beta gene determines susceptibility to nonsense-mediated decay. <i>Human Genetics</i> , <b>2005</b> , 118, 214-24	6.3	42
198	Genetic variations in the gene encoding TFAP2B are associated with type 2 diabetes mellitus. Journal of Human Genetics, <b>2005</b> , 50, 283-292	4.3	63
197	Microalbuminuria as a screening tool in cystic fibrosis-related diabetes. <i>Pediatric Pulmonology</i> , <b>2005</b> , 39, 103-7	3.5	19
196	Relapsing diabetes can result from moderately activating mutations in KCNJ11. <i>Human Molecular Genetics</i> , <b>2005</b> , 14, 925-34	5.6	165
195	A large-scale association analysis of common variation of the HNF1alpha gene with type 2 diabetes in the U.K. Caucasian population. <i>Diabetes</i> , <b>2005</b> , 54, 2487-91	0.9	47
194	High-dose glibenclamide can replace insulin therapy despite transitory diarrhea in early-onset diabetes caused by a novel R201L Kir6.2 mutation. <i>Diabetes Care</i> , <b>2005</b> , 28, 758-9	14.6	75
193	Activating mutations in Kir6.2 and neonatal diabetes: new clinical syndromes, new scientific insights, and new therapy. <i>Diabetes</i> , <b>2005</b> , 54, 2503-13	0.9	356
192	Molecular genetics goes to the diabetes clinic. <i>Clinical Medicine</i> , <b>2005</b> , 5, 476-81	1.9	40
191	Insights into the structure and regulation of glucokinase from a novel mutation (V62M), which causes maturity-onset diabetes of the young. <i>Journal of Biological Chemistry</i> , <b>2005</b> , 280, 14105-13	5.4	76
190	Genetic regulation of birth weight and fasting glucose by a common polymorphism in the islet cell promoter of the glucokinase gene. <i>Diabetes</i> , <b>2005</b> , 54, 576-81	0.9	96
189	Beta-cell dysfunction, insulin sensitivity, and glycosuria precede diabetes in hepatocyte nuclear factor-1alpha mutation carriers. <i>Diabetes Care</i> , <b>2005</b> , 28, 1751-6	14.6	89

#### (2004-2005)

188	Genetic testing for glucokinase mutations in clinically selected patients with MODY: a worthwhile investment. <i>Swiss Medical Weekly</i> , <b>2005</b> , 135, 352-6	3.1	26
187	Lack of support for a role of the insulin gene variable number of tandem repeats minisatellite (INS-VNTR) locus in fetal growth or type 2 diabetes-related intermediate traits in United Kingdom populations. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 310-7	5.6	41
186	Permanent neonatal diabetes due to mutations in KCNJ11 encoding Kir6.2: patient characteristics and initial response to sulfonylurea therapy. <i>Diabetes</i> , <b>2004</b> , 53, 2713-8	0.9	314
185	A simple pragmatic system for detecting new cases of type 2 diabetes and impaired fasting glycaemia in primary care. <i>Family Practice</i> , <b>2004</b> , 21, 57-62	1.9	23
184	Weight differences in Plymouth toddlers compared to the British Growth Reference Population. <i>Archives of Disease in Childhood</i> , <b>2004</b> , 89, 843-4	2.2	5
183	Type 2 diabetes in grandparents and birth weight in offspring and grandchildren in the ALSPAC study. <i>Journal of Epidemiology and Community Health</i> , <b>2004</b> , 58, 517-22	5.1	39
182	Activating mutations in the KCNJ11 gene encoding the ATP-sensitive K+ channel subunit Kir6.2 are rare in clinically defined type 1 diabetes diagnosed before 2 years. <i>Diabetes</i> , <b>2004</b> , 53, 2998-3001	0.9	46
181	Mesangiocapillary glomerulonephritis type 2 associated with familial partial lipodystrophy (Dunnigan-Kobberling syndrome). <i>Nephron Clinical Practice</i> , <b>2004</b> , 96, c35-8		17
180	Common variants of the hepatocyte nuclear factor-4alpha P2 promoter are associated with type 2 diabetes in the U.K. population. <i>Diabetes</i> , <b>2004</b> , 53, 3002-6	0.9	84
179	Molecular basis of Kir6.2 mutations associated with neonatal diabetes or neonatal diabetes plus neurological features. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2004</b> , 101, 17539-44	11.5	205
178	NdonN feel like a diabetic any moreNthe impact of stopping insulin in patients with maturity onset diabetes of the young following genetic testing. <i>Clinical Medicine</i> , <b>2004</b> , 4, 144-7	1.9	46
177	Permanent neonatal diabetes due to paternal germline mosaicism for an activating mutation of the KCNJ11 Gene encoding the Kir6.2 subunit of the beta-cell potassium adenosine triphosphate channel. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2004</b> , 89, 3932-5	5.6	82
176	Messenger RNA transcripts of the hepatocyte nuclear factor-1alpha gene containing premature termination codons are subject to nonsense-mediated decay. <i>Diabetes</i> , <b>2004</b> , 53, 500-4	0.9	44
175	Kir6.2 mutations are a common cause of permanent neonatal diabetes in a large cohort of French patients. <i>Diabetes</i> , <b>2004</b> , 53, 2719-22	0.9	158
174	Evidence from a large U.K. family collection that genes influencing age of onset of type 2 diabetes map to chromosome 12p and to the MODY3/NIDDM2 locus on 12q24. <i>Diabetes</i> , <b>2004</b> , 53, 855-60	0.9	38
173	First UK survey of paediatric type 2 diabetes and MODY. Archives of Disease in Childhood, <b>2004</b> , 89, 526	-92.2	137
172	Contrasting diabetes phenotypes associated with hepatocyte nuclear factor-1alpha and -1beta mutations. <i>Diabetes Care</i> , <b>2004</b> , 27, 1102-7	14.6	99
171	Serum amino acids in patients with mutations in the hepatocyte nuclear factor-1 alpha gene. <i>Diabetic Medicine</i> , <b>2004</b> , 21, 928-30	3.5	6

170	Conventional measures underestimate glycaemia in cystic fibrosis patients. <i>Diabetic Medicine</i> , <b>2004</b> , 21, 691-6	3.5	77
169	The accuracy of birth weight. <i>Journal of Clinical Nursing</i> , <b>2004</b> , 13, 767-8	3.2	4
168	Tall stories: the fundamental difficulties of genetic association studies. <i>Clinical Endocrinology</i> , <b>2004</b> , 60, 145-6	3.4	
167	Premature birth and low birth weight associated with nonautoimmune hyperthyroidism due to an activating thyrotropin receptor gene mutation. <i>Clinical Endocrinology</i> , <b>2004</b> , 60, 711-8	3.4	41
166	Mutations in PTF1A cause pancreatic and cerebellar agenesis. <i>Nature Genetics</i> , <b>2004</b> , 36, 1301-5	36.3	356
165	Heritability estimates for beta cell function and features of the insulin resistance syndrome in UK families with an increased susceptibility to type 2 diabetes. <i>Diabetologia</i> , <b>2004</b> , 47, 732-8	10.3	95
164	Abnormal splicing of hepatocyte nuclear factor-1 beta in the renal cysts and diabetes syndrome. <i>Diabetologia</i> , <b>2004</b> , 47, 937-42	10.3	30
163	Role of the D76N polymorphism of insulin promoter factor-1 in predisposing to Type 2 diabetes. <i>Diabetologia</i> , <b>2004</b> , 47, 957-8	10.3	6
162	Analysis of the contribution to type 2 diabetes susceptibility of sequence variation in the gene encoding stearoyl-CoA desaturase, a key regulator of lipid and carbohydrate metabolism. <i>Diabetologia</i> , <b>2004</b> , 47, 2168-75	10.3	22
161	Association studies of insulin receptor substrate 1 gene (IRS1) variants in type 2 diabetes samples enriched for family history and early age of onset. <i>Diabetes</i> , <b>2004</b> , 53, 3319-22	0.9	36
160	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. <i>New England Journal of Medicine</i> , <b>2004</b> , 350, 1838-49	59.2	930
159	Unlocking the secrets of the pancreatic beta cell: man and mouse provide the key. <i>Journal of Clinical Investigation</i> , <b>2004</b> , 114, 314-6	15.9	8
158	Understanding cystic-fibrosis-related diabetes: best thought of as insulin deficiency?. <i>Journal of the Royal Society of Medicine</i> , <b>2004</b> , 97 Suppl 44, 26-35	2.3	4
157	Genetic variation in the small heterodimer partner gene and young-onset type 2 diabetes, obesity, and birth weight in U.K. subjects. <i>Diabetes</i> , <b>2003</b> , 52, 1276-9	0.9	39
156	Validation of interstitial fluid continuous glucose monitoring in cystic fibrosis. <i>Diabetes Care</i> , <b>2003</b> , 26, 1940-1	14.6	32
155	Etiological investigation of diabetes in young adults presenting with apparent type 2 diabetes. <i>Diabetes Care</i> , <b>2003</b> , 26, 2088-93	14.6	50
154	Apolipoprotein-e influences aspects of intellectual ability in type 1 diabetes. <i>Diabetes</i> , <b>2003</b> , 52, 145-8	0.9	19
153	A genome-wide scan in families with maturity-onset diabetes of the young: evidence for further genetic heterogeneity. <i>Diabetes</i> , <b>2003</b> , 52, 872-81	0.9	55

## (2003-2003)

152	Dimensions of Personal Loss and Gain Associated with a Rare Genetic Type of Diabetes. <i>Illness Crisis and Loss</i> , <b>2003</b> , 11, 362-376	0.6	4	
151	Quantitative traits associated with the Type 2 diabetes susceptibility allele in Kir6.2. <i>Diabetologia</i> , <b>2003</b> , 46, 1021-3	10.3	13	
150	Identification of 21 novel glucokinase (GCK) mutations in UK and European Caucasians with maturity-onset diabetes of the young (MODY). <i>Human Mutation</i> , <b>2003</b> , 22, 417	4.7	64	
149	Atypical familial juvenile hyperuricemic nephropathy associated with a hepatocyte nuclear factor-1beta gene mutation. <i>Kidney International</i> , <b>2003</b> , 63, 1645-51	9.9	120	
148	Adolescent onset Type 2 diabetes in a non-obese Caucasian patient with an unbalanced translocation. <i>Diabetic Medicine</i> , <b>2003</b> , 20, 483-5	3.5	7	
147	Response to treatment with rosiglitazone in familial partial lipodystrophy due to a mutation in the LMNA gene. <i>Diabetic Medicine</i> , <b>2003</b> , 20, 823-7	3.5	53	
146	Variants in the aromatase gene and on the Y-chromosome are not associated with adult height or insulin resistance in a UK population. <i>Clinical Endocrinology</i> , <b>2003</b> , 59, 175-9	3.4	8	
145	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	
144	Association and haplotype analysis of the insulin-degrading enzyme (IDE) gene, a strong positional and biological candidate for type 2 diabetes susceptibility. <i>Diabetes</i> , <b>2003</b> , 52, 1300-5	0.9	49	
143	No deterioration in glycemic control in HNF-1alpha maturity-onset diabetes of the young following transfer from long-term insulin to sulphonylureas. <i>Diabetes Care</i> , <b>2003</b> , 26, 3191-2	14.6	98	
142	How well do midwives estimate the date of delivery?. <i>Midwifery</i> , <b>2003</b> , 19, 125-31	2.8	3	
141	Apolipoprotein E genotype, islet amyloid deposition and severity of Type 2 diabetes. <i>Diabetes Research and Clinical Practice</i> , <b>2003</b> , 60, 105-10	7-4	13	
140	Genetic cause of hyperglycaemia and response to treatment in diabetes. <i>Lancet, The</i> , <b>2003</b> , 362, 1275-8	140	437	
139	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	
138	Regulation of apolipoprotein M gene expression by MODY3 gene hepatocyte nuclear factor-1alpha: haploinsufficiency is associated with reduced serum apolipoprotein M levels. <i>Diabetes</i> , <b>2003</b> , 52, 2989-95	0.9	108	
137	Young-onset type 2 diabetes families are the major contributors to genetic loci in the Diabetes UK Warren 2 genome scan and identify putative novel loci on chromosomes 8q21, 21q22, and 22q11. <i>Diabetes</i> , <b>2003</b> , 52, 1857-63	0.9	38	
136	Insights into the biochemical and genetic basis of glucokinase activation from naturally occurring hypoglycemia mutations. <i>Diabetes</i> , <b>2003</b> , 52, 2433-40	0.9	133	
135	Identifying hepatic nuclear factor 1alpha mutations in children and young adults with a clinical diagnosis of type 1 diabetes. <i>Diabetes Care</i> , <b>2003</b> , 26, 333-7	14.6	74	

134	A meta-analysis of four European genome screens (GIFT Consortium) shows evidence for a novel region on chromosome 17p11.2-q22 linked to type 2 diabetes. <i>Human Molecular Genetics</i> , <b>2003</b> , 12, 18	65 <sup>5</sup> 73	60
133	Distinct molecular and morphogenetic properties of mutations in the human HNF1beta gene that lead to defective kidney development. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2003</b> , 14, 2033-41	12.7	68
132	Severe hyperglycemia after renal transplantation in a pediatric patient with a mutation of the hepatocyte nuclear factor-1beta gene. <i>American Journal of Kidney Diseases</i> , <b>2002</b> , 40, 1325-30	7.4	17
131	Complete glucokinase deficiency is not a common cause of permanent neonatal diabetes. <i>Diabetologia</i> , <b>2002</b> , 45, 290	10.3	25
130	The genetic abnormality in the beta cell determines the response to an oral glucose load. <i>Diabetologia</i> , <b>2002</b> , 45, 427-35	10.3	197
129	Human calcium/calmodulin-dependent protein kinase II gamma gene (CAMK2G): cloning, genomic structure and detection of variants in subjects with type II diabetes. <i>Diabetologia</i> , <b>2002</b> , 45, 580-3	10.3	14
128	Association of calpain-10 gene with microvascular function. <i>Diabetologia</i> , <b>2002</b> , 45, 899-904	10.3	23
127	Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1beta mutations. <i>Kidney International</i> , <b>2002</b> , 61, 1243-51	9.9	104
126	Heterogeneity in young adult onset diabetes: aetiology alters clinical characteristics. <i>Diabetic Medicine</i> , <b>2002</b> , 19, 758-61	3.5	38
125	Rare variants identified in the HNF- 4 alpha beta-cell-specific promoter and alternative exon 1 lack biological significance in maturity onset diabetes of the young and young onset Type II diabetes. <i>Diabetologia</i> , <b>2002</b> , 45, 1344-8	10.3	16
124	Abnormal splicing of hepatocyte nuclear factor 1 alpha in maturity-onset diabetes of the young. <i>Diabetologia</i> , <b>2002</b> , 45, 1463-7	10.3	19
123	Conflicting results on variation in the IGFI gene highlight methodological considerations in the design of genetic association studies. <i>Diabetologia</i> , <b>2002</b> , 45, 1605-6	10.3	11
122	Variation in the calpain-10 gene affects blood glucose levels in the British population. <i>Diabetes</i> , <b>2002</b> , 51, 247-50	0.9	71
121	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. <i>Diabetes</i> , <b>2002</b> , 51, 2313-6	0.9	123
120	Necrobiosis lipodica is a clinical feature of maturity-onset diabetes of the young. <i>Diabetes Care</i> , <b>2002</b> , 25, 1249-50	14.6	6
119	Evidence for haploinsufficiency of the human HNF1alpha gene revealed by functional characterization of MODY3-associated mutations. <i>Biological Chemistry</i> , <b>2002</b> , 383, 1691-700	4.5	23
118	Association studies of genetic variation in the WFS1 gene and type 2 diabetes in U.K. populations. <i>Diabetes</i> , <b>2002</b> , 51, 1287-90	0.9	79
117	The role of genetic susceptibility in diabetic nephropathy: evidence from family studies. <i>Nephrology Dialysis Transplantation</i> , <b>2002</b> , 17, 1543-6	4.3	22

## (2001-2002)

116	Variation within the type 2 diabetes susceptibility gene calpain-10 and polycystic ovary syndrome. Journal of Clinical Endocrinology and Metabolism, <b>2002</b> , 87, 2606-10	5.6	57
115	Increased risk of diabetes in first-degree relatives of young-onset type 2 diabetic patients compared with relatives of those diagnosed later. <i>Diabetes Care</i> , <b>2002</b> , 25, 636-7	14.6	16
114	Clinical improvement in cystic fibrosis with early insulin treatment. <i>Archives of Disease in Childhood</i> , <b>2002</b> , 87, 430-1	2.2	89
113	Maturity-onset diabetes of the young caused by a balanced translocation where the 20q12 break point results in disruption upstream of the coding region of hepatocyte nuclear factor-4alpha (HNF4A) gene. <i>Diabetes</i> , <b>2002</b> , 51, 2329-33	0.9	25
112	Different genes, different diabetes: lessons from maturity-onset diabetes of the young. <i>Annals of Medicine</i> , <b>2002</b> , 34, 207-216	1.5	138
111	Frequent occurrence of an intron 4 mutation in multiple endocrine neoplasia type 1. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2002</b> , 87, 2688-93	5.6	47
110	Intrauterine hyperglycemia is associated with an earlier diagnosis of diabetes in HNF-1alpha gene mutation carriers. <i>Diabetes Care</i> , <b>2002</b> , 25, 2287-91	14.6	84
109	Evidence for linkage of stature to chromosome 3p26 in a large U.K. Family data set ascertained for type 2 diabetes. <i>American Journal of Human Genetics</i> , <b>2002</b> , 70, 543-6	11	44
108	The role of the HNF4alpha enhancer in type 2 diabetes. <i>Molecular Genetics and Metabolism</i> , <b>2002</b> , 76, 148-51	3.7	14
107	Different genes, different diabetes: lessons from maturity-onset diabetes of the young. <i>Annals of Medicine</i> , <b>2002</b> , 34, 207-16	1.5	37
106	Predictive genetic testing in maturity-onset diabetes of the young (MODY). <i>Diabetic Medicine</i> , <b>2001</b> , 18, 417-21	3.5	49
105	Skin microvascular vasodilatory capacity in offspring of two parents with Type 2 diabetes. <i>Diabetic Medicine</i> , <b>2001</b> , 18, 541-5	3.5	13
104	Glucokinase mutations in a phenotypically selected multiethnic group of women with a history of gestational diabetes. <i>Diabetic Medicine</i> , <b>2001</b> , 18, 683-4	3.5	28
103	Influence of maternal and fetal glucokinase mutations in gestational diabetes. <i>American Journal of Obstetrics and Gynecology</i> , <b>2001</b> , 185, 240-1	6.4	92
102	The role of genetic susceptibility in the association of low birth weight with type 2 diabetes. <i>British Medical Bulletin</i> , <b>2001</b> , 60, 89-101	5.4	50
101	A distant upstream promoter of the HNF-4alpha gene connects the transcription factors involved in maturity-onset diabetes of the young. <i>Human Molecular Genetics</i> , <b>2001</b> , 10, 2089-97	5.6	158
100	Kidney disease in hypomelanosis of Ito. Nephrology Dialysis Transplantation, 2001, 16, 1267-9	4.3	11
99	Adult height and proteinuria in type 2 diabetes. <i>Nephrology Dialysis Transplantation</i> , <b>2001</b> , 16, 525-8	4.3	10

98	Molecular diagnostics in monogenic and multifactorial forms of type 2 diabetes. <i>Expert Review of Molecular Diagnostics</i> , <b>2001</b> , 1, 403-12	3.8	35
97	The generalized aminoaciduria seen in patients with hepatocyte nuclear factor-1alpha mutations is a feature of all patients with diabetes and is associated with glucosuria. <i>Diabetes</i> , <b>2001</b> , 50, 2047-52	0.9	50
96	ACE gene polymorphism as a prognostic indicator in patients with type 2 diabetes and established renal disease. <i>Diabetes Care</i> , <b>2001</b> , 24, 2115-20	14.6	29
95	beta-cell genes and diabetes: molecular and clinical characterization of mutations in transcription factors. <i>Diabetes</i> , <b>2001</b> , 50 Suppl 1, S94-100	0.9	203
94	Maturity-onset diabetes of the young: from clinical description to molecular genetic characterization. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , <b>2001</b> , 15, 309-23	6.5	103
93	Mutations in the hepatocyte nuclear factor-1beta gene are associated with familial hypoplastic glomerulocystic kidney disease. <i>American Journal of Human Genetics</i> , <b>2001</b> , 68, 219-24	11	227
92	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
91	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
90	Development of a bloodspot assay for insulin. <i>Clinica Chimica Acta</i> , <b>2001</b> , 310, 141-50	6.2	21
89	beta-cell genes and diabetes: quantitative and qualitative differences in the pathophysiology of hepatic nuclear factor-1alpha and glucokinase mutations. <i>Diabetes</i> , <b>2001</b> , 50 Suppl 1, S101-7	0.9	103
88	Hepatocyte nuclear factor-1beta: a new kindred with renal cysts and diabetes and gene expression in normal human development. <i>Journal of the American Society of Nephrology: JASN</i> , <b>2001</b> , 12, 2175-218	зб <sup>2.7</sup>	84
87	Clinical Consequences of Defects in Ecell Genes. <i>Growth Hormone</i> , <b>2001</b> , 325-336		
86	Proposed mechanism for a novel insertion/deletion frameshift mutation (I414G415ATCG>CCA) in the hepatocyte nuclear factor 1 alpha (HNF-1 alpha) gene which causes maturity-onset diabetes of the young (MODY). <i>Human Mutation</i> , <b>2000</b> , 16, 273	4.7	8
85	Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1 beta. <i>Kidney International</i> , <b>2000</b> , 57, 898-907	9.9	132
84	Sensitivity to sulphonylureas in patients with hepatocyte nuclear factor-1alpha gene mutations: evidence for pharmacogenetics in diabetes. <i>Diabetic Medicine</i> , <b>2000</b> , 17, 543-5	3.5	198
83	Heterogeneity in the clinical course of patients with Type 2 diabetes on dialysisthe need for different preventative strategies. <i>Diabetic Medicine</i> , <b>2000</b> , 17, 685-6	3.5	2
82	Mutations in the human delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. <i>Nature Genetics</i> , <b>2000</b> , 24, 438-41	36.3	308
81	Intermediate expansions of a X25/frataxin gene GAA repeat and type II diabetes: assessment using parent-offspring trios. <i>Diabetologia</i> , <b>2000</b> , 43, 384-5	10.3	5

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80	Maternal diabetes alters birth weight in glucokinase-deficient (MODY2) kindred but has no influence on adult weight, height, insulin secretion or insulin sensitivity. <i>Diabetologia</i> , <b>2000</b> , 43, 1060-3	10.3	61
79	R127W in HNF4alpha is a loss-of-function mutation causing maturity-onset diabetes of the young (MODY) in a UK Caucasian family. <i>Diabetologia</i> , <b>2000</b> , 43, 1203	10.3	4
78	Evidence that single nucleotide polymorphism in the uncoupling protein 3 (UCP3) gene influences fat distribution in women of European and Asian origin. <i>Diabetologia</i> , <b>2000</b> , 43, 1558-64	10.3	71
77	A high prevalence of glucokinase mutations in gestational diabetic subjects selected by clinical criteria. <i>Diabetologia</i> , <b>2000</b> , 43, 250-3	10.3	158
76	Diagnosis of maturity-onset diabetes of the young in the pediatric diabetes clinic. <i>Journal of Pediatric Endocrinology and Metabolism</i> , <b>2000</b> , 13 Suppl 6, 1411-7	1.6	6
75	The mutated human gene encoding hepatocyte nuclear factor 1beta inhibits kidney formation in developing Xenopus embryos. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2000</b> , 97, 4695-700	11.5	69
74	No evidence for linkage at candidate type 2 diabetes susceptibility loci on chromosomes 12 and 20 in United Kingdom Caucasians. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2000</b> , 85, 853-7	5.6	15
73	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. <i>Diabetes</i> , <b>2000</b> , 49, 126-30	0.9	159
72	Predictive genetic testing in diabetes: a case study of multiple perspectives. <i>Qualitative Health Research</i> , <b>2000</b> , 10, 242-59	3.9	24
71	Naturally occurring mutations in the human HNF4alpha gene impair the function of the transcription factor to a varying degree. <i>Nucleic Acids Research</i> , <b>2000</b> , 28, 430-7	20.1	58
70	A severe clinical phenotype results from the co-inheritance of type 2 susceptibility genes and a hepatocyte nuclear factor-1alpha mutation. <i>Diabetes Care</i> , <b>2000</b> , 23, 424-5	14.6	12
69	The HOPE study and diabetes. Heart Outcomes Prevention Evaluation. <i>Lancet, The</i> , <b>2000</b> , 355, 1182-3; author reply 1183-4	40	6
68	Increased prevalence of proteinuria in diabetic sibs of proteinuric type 2 diabetic subjects. <i>American Journal of Kidney Diseases</i> , <b>2000</b> , 35, 708-12	7.4	23
67	Searching for type 2 diabetes genes in the post-genome era. <i>Trends in Endocrinology and Metabolism</i> , <b>2000</b> , 11, 383-93	8.8	34
66	Hypoglycaemic counter-regulation at normal blood glucose concentrations in patients with well controlled type-2 diabetes. <i>Lancet, The</i> , <b>2000</b> , 356, 1970-4	40	93
65	Parent-offspring trios: a resource to facilitate the identification of type 2 diabetes genes. <i>Diabetes</i> , <b>1999</b> , 48, 2475-9	0.9	39
64	Impaired maximum microvascular hyperaemia in patients with MODY 3 (hepatocyte nuclear factor-1alpha gene mutations). <i>Diabetic Medicine</i> , <b>1999</b> , 16, 731-5	3.5	7
63	Non-penetrance in a MODY 3 family with a mutation in the hepatic nuclear factor 1alpha gene: implications for predictive testing. <i>European Journal of Human Genetics</i> , <b>1999</b> , 7, 729-32	5.3	12

62	Loss of HNF1[function in human renal cell carcinoma: Frequent mutations in the VHL gene but not the HNF1[gene <b>1999</b> , 24, 305-314		11
61	Allelic drop-out in exon 2 of the hepatocyte nuclear factor-1alpha gene hinders the identification of mutations in three families with maturity-onset diabetes of the young. <i>Diabetes</i> , <b>1999</b> , 48, 921-3	0.9	26
60	A gene for autosomal recessive spondylocostal dysostosis maps to 19q13.1-q13.3. <i>American Journal of Human Genetics</i> , <b>1999</b> , 65, 175-82	11	65
59	Pigmentary retinal dystrophy and the syndrome of maternally inherited diabetes and deafness caused by the mitochondrial DNA 3243 tRNA(Leu) A to G mutation. <i>Ophthalmology</i> , <b>1999</b> , 106, 1101-8	7.3	69
58	The fetal insulin hypothesis: an alternative explanation of the association of low birthweight with diabetes and vascular disease. <i>Lancet, The</i> , <b>1999</b> , 353, 1789-92	40	729
57	Missense mutations in the insulin promoter factor-1 gene predispose to type 2 diabetes. <i>Journal of Clinical Investigation</i> , <b>1999</b> , 104, R33-9	15.9	176
56	Loss of HNF1alpha function in human renal cell carcinoma: frequent mutations in the VHL gene but not the HNF1alpha gene. <i>Molecular Carcinogenesis</i> , <b>1999</b> , 24, 305-14	5	7
55	Mutations in the glucokinase gene of the fetus result in reduced birth weight. <i>Nature Genetics</i> , <b>1998</b> , 19, 268-70	36.3	483
54	Maturity-onset diabetes of the young: clinical heterogeneity explained by genetic heterogeneity. <i>Diabetic Medicine</i> , <b>1998</b> , 15, 15-24	3.5	184
53	Mutations in hepatocyte nuclear factor 1beta are not a common cause of maturity-onset diabetes of the young in the U.K. <i>Diabetes</i> , <b>1998</b> , 47, 1152-4	0.9	46
52	Mutations in the hepatocyte nuclear factor-1alpha gene are a common cause of maturity-onset diabetes of the young in the U.K. <i>Diabetes</i> , <b>1997</b> , 46, 720-5	0.9	138
51	Genes versus environment in insulin-dependent diabetes: the phoney war. <i>Lancet, The</i> , <b>1997</b> , 349, 147-8	340	12
50	Genetic Factors in the Aetiology of Non-Insulin-Dependent Diabetes. <i>Frontiers of Hormone Research</i> , <b>1997</b> , 22, 157-178	3.5	1
49	A rapid screening method for hepatocyte nuclear factor 1 alpha frameshift mutations; prevalence in maturity-onset diabetes of the young and late-onset non-insulin dependent diabetes. <i>Human Genetics</i> , <b>1997</b> , 101, 351-4	6.3	26
48	A missense mutation in the hepatocyte nuclear factor 4 alpha gene in a UK pedigree with maturity-onset diabetes of the young. <i>Diabetologia</i> , <b>1997</b> , 40, 859-62	10.3	55
47	Glucokinase deficiency results in a beta-cell disorder characterised by normal fasting plasma proinsulin concentrations. <i>Diabetologia</i> , <b>1997</b> , 40, 1367-8	10.3	8
46	UKPDS 21: low prevalence of the mitochondrial transfer RNA gene (tRNA(Leu(UUR))) mutation at position 3243bp in UK Caucasian type 2 diabetic patients. <i>Diabetic Medicine</i> , <b>1997</b> , 14, 42-5	3.5	33
45	The mitochondrial tRNA[Leu(UUR)] A to G 3243 mutation is associated with insulin-dependent and non-insulin-dependent diabetes in a Chinese population. <i>Diabetic Medicine</i> , <b>1997</b> , 14, 1026-31	3.5	14

44	Mutations in the hepatocyte nuclear factor-1alpha gene are a common cause of maturity-onset diabetes of the young in the U.K. <i>Diabetes</i> , <b>1997</b> , 46, 720-725	0.9	45
43	High prevalence of a missense mutation of the glucokinase gene in gestational diabetic patients due to a founder-effect in a local population. <i>Diabetologia</i> , <b>1996</b> , 39, 1325-8	10.3	53
42	Mutations in the hepatocyte nuclear factor-1alpha gene in maturity-onset diabetes of the young (MODY3). <i>Nature</i> , <b>1996</b> , 384, 455-8	50.4	973
41	Altered insulin secretory responses to glucose in diabetic and nondiabetic subjects with mutations in the diabetes susceptibility gene MODY3 on chromosome 12. <i>Diabetes</i> , <b>1996</b> , 45, 1503-10	0.9	216
40	Altered insulin secretory responses to glucose in diabetic and nondiabetic subjects with mutations in the diabetes susceptibility gene MODY3 on chromosome 12. <i>Diabetes</i> , <b>1996</b> , 45, 1503-1510	0.9	55
39	Prevalence of diabetes mellitus and impaired glucose tolerance in parents of women with gestational diabetes. <i>Diabetologia</i> , <b>1995</b> , 38, 693-8	10.3	26
38	Candidate gene studies in pedigrees with maturity-onset diabetes of the young not linked with glucokinase. <i>Diabetologia</i> , <b>1995</b> , 38, 1055-60	10.3	15
37	Clinical characteristics of subjects with a missense mutation in glucokinase. <i>Diabetic Medicine</i> , <b>1995</b> , 12, 209-17	3.5	52
36	Type II diabetes: clinical aspects of molecular biological studies. <i>Diabetes</i> , <b>1995</b> , 44, 1-10	0.9	66
35	Type II diabetes: clinical aspects of molecular biological studies. <i>Diabetes</i> , <b>1995</b> , 44, 1-10	0.9	22
34	Candidate gene studies in pedigrees with maturity-onset diabetes of the young not linked with glucokinase <b>1995</b> , 38, 1055		1
33	The effect of long- and short-term corticosteroids on plasma calcitonin and parathyroid hormone levels. <i>Calcified Tissue International</i> , <b>1994</b> , 54, 198-202	3.9	42
32	Segregation analysis of NIDDM in Caucasian families. <i>Diabetologia</i> , <b>1994</b> , 37, 1231-40	10.3	30
31	Non-linkage of the glucagon-like peptide 1 receptor gene with maturity onset diabetes of the young. <i>Diabetologia</i> , <b>1994</b> , 37, 721-4	10.3	31
30	Non-linkage of the glucagon-like peptide 1 receptor gene with maturity onset diabetes of the young <b>1994</b> , 37, 721		4
29	Segregation analysis of NIDDM in Caucasian families <b>1994</b> , 37, 1231		2
28	Distribution of type II diabetes in nuclear families. <i>Diabetes</i> , <b>1993</b> , 42, 106-12	0.9	27
27	Availability of type II diabetic families for detection of diabetes susceptibility genes. <i>Diabetes</i> , <b>1993</b> , 42, 1536-43	0.9	20

26	Microsatellite polymorphisms at the glucokinase locus: a population association study in Caucasian type 2 diabetic subjects. <i>Diabetic Medicine</i> , <b>1993</b> , 10, 694-8	3.5	5
25	Linkage of maturity-onset diabetes of the young to the glucokinase geneevidence of genetic heterogeneity. <i>Biochemical Society Transactions</i> , <b>1993</b> , 21, 24S	5.1	1
24	Association of low birth weight with beta cell function in the adult first degree relatives of non-insulin dependent diabetic subjects. <i>BMJ: British Medical Journal</i> , <b>1993</b> , 306, 302-6		60
23	Distribution of type II diabetes in nuclear families. <i>Diabetes</i> , <b>1993</b> , 42, 106-112	0.9	2
22	Availability of type II diabetic families for detection of diabetes susceptibility genes. <i>Diabetes</i> , <b>1993</b> , 42, 1536-1543	0.9	2
21	Linkage analysis of glucokinase gene with NIDDM in Caucasian pedigrees. <i>Diabetes</i> , <b>1992</b> , 41, 1496-500	0.9	34
20	Type II diabetes: search for primary defects. <i>Annals of Medicine</i> , <b>1992</b> , 24, 511-6	1.5	8
19	Beta-cell secretory defect caused by mutations in glucokinase gene. <i>Lancet, The</i> , <b>1992</b> , 340, 1162-3	40	10
18	Linkage of type 2 diabetes to the glucokinase gene. <i>Lancet, The</i> , <b>1992</b> , 339, 1307-10	40	343
17	Failure to detect cytomegalovirus DNA in pancreas in type 2 diabetes. <i>Lancet, The</i> , <b>1992</b> , 339, 459-60	40	11
16	A survey of cytomegalovirus (CMV) DNA in primary sclerosing cholangitis (PSC) liver tissues using a sensitive polymerase chain reaction (PCR) based assay. <i>Journal of Hepatology</i> , <b>1992</b> , 15, 396-9	13.4	28
15	Missense glucokinase mutation in maturity-onset diabetes of the young and mutation screening in late-onset diabetes. <i>Nature Genetics</i> , <b>1992</b> , 2, 153-6	36.3	131
14	Linkage analysis of glucokinase gene with NIDDM in Caucasian pedigrees. <i>Diabetes</i> , <b>1992</b> , 41, 1496-1500	00.9	9
13	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. <i>Wellcome Open Research</i> ,5, 175	4.8	
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