Sian Ellard

List of Publications by Citations

490 papers	30,621 citations	88 h-index	160 g-index
518 ext. papers	34,949 ext. citations	6.9 avg, IF	6.66 L-index

#	Paper	IF	Citations
490	A common variant in the FTO gene is associated with body mass index and predisposes to childhood and adult obesity. *Science*, **2007**, 316, 889-94	33.3	3294
489	Replication of genome-wide association signals in UK samples reveals risk loci for type 2 diabetes. *Science*, **2007**, 316, 1336-41	33.3	1823
488	Activating mutations in the gene encoding the ATP-sensitive potassium-channel subunit Kir6.2 and permanent neonatal diabetes. *New England Journal of Medicine*, **2004**, 350, 1838-49	59.2	930
487	Switching from insulin to oral sulfonylureas in patients with diabetes due to Kir6.2 mutations. *New England Journal of Medicine*, **2006**, 355, 467-77	59.2	740
486	Mutations in the glucokinase gene of the fetus result in reduced birth weight. *Nature Genetics*, **1998**, 19, 268-70	36.3	483
485	Maturity-onset diabetes of the young (MODY): how many cases are we missing?. *Diabetologia*, **2010** , 53, 2504-8	10.3	432
484	Insulin gene mutations as a cause of permanent neonatal diabetes. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 15040-4	11.5	426
483	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. *Nature Genetics*, **2008**, 40, 949-51	36.3	417
482	Clinical implications of a molecular genetic classification of monogenic beta-cell diabetes. *Nature Clinical Practice Endocrinology and Metabolism*, **2008**, 4, 200-13		367
481	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. *Nature Genetics*, **2014**, 46, 812-814	36.3	328
480	Update on mutations in glucokinase (GCK), which cause maturity-onset diabetes of the young, permanent neonatal diabetes, and hyperinsulinemic hypoglycemia. *Human Mutation*, **2009**, 30, 1512-26	4.7	324
479	Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. *Diabetologia*, **2008**, 51, 546-53	10.3	311
478	Mutations in the human delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. *Nature Genetics*, **2000**, 24, 438-41	36.3	308
477	Insulin mutation screening in 1,044 patients with diabetes: mutations in the INS gene are a common cause of neonatal diabetes but a rare cause of diabetes diagnosed in childhood or adulthood. *Diabetes*, **2008**, 57, 1034-42	0.9	299
476	Macrosomia and hyperinsulinaemic hypoglycaemia in patients with heterozygous mutations in the HNF4A gene. *PLoS Medicine*, **2007**, 4, e118	11.6	279
475	Mutations in ATP-sensitive K+ channel genes cause transient neonatal diabetes and permanent diabetes in childhood or adulthood. *Diabetes*, **2007**, 56, 1930-7	0.9	273
474	Using SIFT and PolyPhen to predict loss-of-function and gain-of-function mutations. *Genetic Testing and Molecular Biomarkers*, **2010**, 14, 533-7	1.6	247

473	Alagille syndrome: pathogenesis, diagnosis and management. *European Journal of Human Genetics*, **2012**, 20, 251-7	5.3	239
472	Mutations in hepatocyte nuclear factor-1beta and their related phenotypes. *Journal of Medical Genetics*, **2006**, 43, 84-90	5.8	236
471	Mutations in the hepatocyte nuclear factor-1beta gene are associated with familial hypoplastic glomerulocystic kidney disease. *American Journal of Human Genetics*, **2001**, 68, 219-24	11	227
470	Prevalence, characteristics and clinical diagnosis of maturity onset diabetes of the young due to mutations in HNF1A, HNF4A, and glucokinase: results from the SEARCH for Diabetes in Youth. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 4055-62	5.6	219
469	Effective treatment with oral sulfonylureas in patients with diabetes due to sulfonylurea receptor 1 (SUR1) mutations. *Diabetes Care*, **2008**, 31, 204-9	14.6	203
468	Improved genetic testing for monogenic diabetes using targeted next-generation sequencing. *Diabetologia*, **2013**, 56, 1958-63	10.3	201
467	Exome sequencing identifies a DYNC1H1 mutation in a large pedigree with dominant axonal Charcot-Marie-Tooth disease. *American Journal of Human Genetics*, **2011**, 89, 308-12	11	199
466	GATA6 haploinsufficiency causes pancreatic agenesis in humans. *Nature Genetics*, **2011**, 44, 20-22	36.3	195
465	HNF1B mutations associate with hypomagnesemia and renal magnesium wasting. *Journal of the American Society of Nephrology: JASN*, **2009**, 20, 1123-31	12.7	192
464	Mutations in KCNJ11, which encodes Kir6.2, are a common cause of diabetes diagnosed in the first 6 months of life, with the phenotype determined by genotype. *Diabetologia*, **2006**, 49, 1190-7	10.3	190
463	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. *Nature Genetics*, **2014**, 46, 61-64	36.3	187
462	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. *Lancet, The*, **2015**, 386, 957-63	40	186
461	Prevalence of vascular complications among patients with glucokinase mutations and prolonged, mild hyperglycemia. *JAMA - Journal of the American Medical Association*, **2014**, 311, 279-86	27.4	181
460	Update of mutations in the genes encoding the pancreatic beta-cell K(ATP) channel subunits Kir6.2 (KCNJ11) and sulfonylurea receptor 1 (ABCC8) in diabetes mellitus and hyperinsulinism. *Human Mutation*, **2009**, 30, 170-80	4.7	181
459	Missense mutations in the insulin promoter factor-1 gene predispose to type 2 diabetes. *Journal of Clinical Investigation*, **1999**, 104, R33-9	15.9	176
458	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. *Human Molecular Genetics*, **2006**, 15, 1793-800	5.6	175
457	The development and validation of a clinical prediction model to determine the probability of MODY in patients with young-onset diabetes. *Diabetologia*, **2012**, 55, 1265-72	10.3	172
456	Molecular genetics and phenotypic characteristics of MODY caused by hepatocyte nuclear factor 4alpha mutations in a large European collection. *Diabetologia*, **2005**, 48, 878-85	10.3	170

455	A genetic diagnosis of HNF1A diabetes alters treatment and improves glycaemic control in the majority of insulin-treated patients. *Diabetic Medicine*, **2009**, 26, 437-41	3.5	168
454	Relapsing diabetes can result from moderately activating mutations in KCNJ11. *Human Molecular Genetics*, **2005**, 14, 925-34	5.6	165
453	HNF1B-associated renal and extra-renal disease-an expanding clinical spectrum. *Nature Reviews Nephrology*, **2015**, 11, 102-12	14.9	163
452	Permanent neonatal diabetes caused by dominant, recessive, or compound heterozygous SUR1 mutations with opposite functional effects. *American Journal of Human Genetics*, **2007**, 81, 375-82	11	161
451	A high prevalence of glucokinase mutations in gestational diabetic subjects selected by clinical criteria. *Diabetologia*, **2000**, 43, 250-3	10.3	158
450	Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation. *Diabetes Care*, **2015**, 38, 1383-92	14.6	157
449	Studies of association between the gene for calpain-10 and type 2 diabetes mellitus in the United Kingdom. *American Journal of Human Genetics*, **2001**, 69, 544-52	11	154
448	Prospective functional classification of all possible missense variants in PPARG. *Nature Genetics*, **2016**, 48, 1570-1575	36.3	149
447	Recessive mutations in the INS gene result in neonatal diabetes through reduced insulin biosynthesis. *Proceedings of the National Academy of Sciences of the United States of America*, **2010**, 107, 3105-10	11.5	149
446	Clinical and molecular characterisation of 300 patients with congenital hyperinsulinism. *European Journal of Endocrinology*, **2013**, 168, 557-64	6.5	147
445	Mutations in the genes encoding the transcription factors hepatocyte nuclear factor 1 alpha (HNF1A) and 4 alpha (HNF4A) in maturity-onset diabetes of the young. *Human Mutation*, **2006**, 27, 854-69	4.7	137
444	Characterization of aryl hydrocarbon receptor interacting protein (AIP) mutations in familial isolated pituitary adenoma families. *Human Mutation*, **2010**, 31, 950-60	4.7	136
443	ISPAD Clinical Practice Consensus Guidelines 2018: The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatric Diabetes*, **2018**, 19 Suppl 27, 47-63	3.6	136
442	ISPAD Clinical Practice Consensus Guidelines 2014. The diagnosis and management of monogenic diabetes in children and adolescents. *Pediatric Diabetes*, **2014**, 15 Suppl 20, 47-64	3.6	133
441	Insights into the biochemical and genetic basis of glucokinase activation from naturally occurring hypoglycemia mutations. *Diabetes*, **2003**, 52, 2433-40	0.9	133
440	Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1 beta. *Kidney International*, **2000**, 57, 898-907	9.9	132
439	Heterogeneous genetic background of the association of pheochromocytoma/paraganglioma and pituitary adenoma: results from a large patient cohort. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E531-41	5.6	127
438	Islet autoantibodies can discriminate maturity-onset diabetes of the young (MODY) from Type 1 diabetes. *Diabetic Medicine*, **2011**, 28, 1028-33	3.5	127

437	Human CHN1 mutations hyperactivate alpha2-chimaerin and cause Duane's retraction syndrome. *Science*, **2008**, 321, 839-43	33.3	126
436	Mutated MESP2 causes spondylocostal dysostosis in humans. *American Journal of Human Genetics*, **2004**, 74, 1249-54	11	124
435	Hepatocyte nuclear factor-1 beta mutations cause neonatal diabetes and intrauterine growth retardation: support for a critical role of HNF-1beta in human pancreatic development. *Diabetic Medicine*, **2006**, 23, 1301-6	3.5	123
434	Cross-sectional and longitudinal studies suggest pharmacological treatment used in patients with glucokinase mutations does not alter glycaemia. *Diabetologia*, **2014**, 57, 54-6	10.3	122
433	Systematic assessment of etiology in adults with a clinical diagnosis of young-onset type 2 diabetes is a successful strategy for identifying maturity-onset diabetes of the young. *Diabetes Care*, **2012**, 35, 1206-12	14.6	122
432	AIP mutation in pituitary adenomas in the 18th century and today. *New England Journal of Medicine* , **2011**, 364, 43-50	59.2	122
431	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. *Nature Genetics*, **2013**, 45, 947-50	36.3	120
430	Atypical familial juvenile hyperuricemic nephropathy associated with a hepatocyte nuclear factor-1beta gene mutation. *Kidney International*, **2003**, 63, 1645-51	9.9	120
429	Mutations in the genes encoding the transcription factors hepatocyte nuclear factor 1 alpha and 4 alpha in maturity-onset diabetes of the young and hyperinsulinemic hypoglycemia. *Human Mutation* , **2013**, 34, 669-85	4.7	119
428	Systematic Population Screening, Using Biomarkers and Genetic Testing, Identifies 2.5% of the U.K. Pediatric Diabetes Population With Monogenic Diabetes. *Diabetes Care*, **2016**, 39, 1879-1888	14.6	117
427	Homozygous mutations in NEUROD1 are responsible for a novel syndrome of permanent neonatal diabetes and neurological abnormalities. *Diabetes*, **2010**, 59, 2326-31	0.9	116
426	Landscape of Familial Isolated and Young-Onset Pituitary Adenomas: Prospective Diagnosis in AIP Mutation Carriers. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1242-54	5.6	115
425	Permanent Neonatal Diabetes and Enteric Anendocrinosis Associated With Biallelic Mutations in NEUROG3. *Diabetes*, **2011**, 60, 1349-53	0.9	114
424	Persistent hyperinsulinemic hypoglycemia and maturity-onset diabetes of the young due to heterozygous HNF4A mutations. *Diabetes*, **2008**, 57, 1659-63	0.9	113
423	KCNJ11 activating mutations are associated with developmental delay, epilepsy and neonatal diabetes syndrome and other neurological features. *European Journal of Human Genetics*, **2006**, 14, 824-30	5.3	110
422	Heterozygous ABCC8 mutations are a cause of MODY. *Diabetologia*, **2012**, 55, 123-7	10.3	105
421	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. *Diabetes*, **2016**, 65, 2094-2099	0.9	105
420	GATA6 mutations cause a broad phenotypic spectrum of diabetes from pancreatic agenesis to adult-onset diabetes without exocrine insufficiency. *Diabetes*, **2013**, 62, 993-7	0.9	104

419	Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1beta mutations. *Kidney International*, **2002**, 61, 1243-51	9.9	104
418	Wolcott-Rallison syndrome is the most common genetic cause of permanent neonatal diabetes in consanguineous families. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 4162-70	5.6	103
417	Analysis of transcription factors key for mouse pancreatic development establishes NKX2-2 and MNX1 mutations as causes of neonatal diabetes in man. *Cell Metabolism*, **2014**, 19, 146-54	24.6	102
416	Isomers of the TCF1 gene encoding hepatocyte nuclear factor-1 alpha show differential expression in the pancreas and define the relationship between mutation position and clinical phenotype in monogenic diabetes. *Human Molecular Genetics*, **2006**, 15, 2216-24	5.6	100
415	Contrasting diabetes phenotypes associated with hepatocyte nuclear factor-1alpha and -1beta mutations. *Diabetes Care*, **2004**, 27, 1102-7	14.6	99
414	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. *Diabetologia*, **2009**, 52, 1683-5	10.3	98
413	No deterioration in glycemic control in HNF-1alpha maturity-onset diabetes of the young following transfer from long-term insulin to sulphonylureas. *Diabetes Care*, **2003**, 26, 3191-2	14.6	98
412	Sirolimus therapy in infants with severe hyperinsulinemic hypoglycemia. *New England Journal of Medicine*, **2014**, 370, 1131-7	59.2	97
411	Hepatocyte nuclear factor 1 alpha (HNF-1 alpha) mutations in maturity-onset diabetes of the young. *Human Mutation*, **2000**, 16, 377-85	4.7	97
410	Maturity onset diabetes of the young: identification and diagnosis. *Annals of Clinical Biochemistry*, **2013**, 50, 403-15	2.2	96
409	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. *Diabetes Care*, **2011**, 34, 286-91	14.6	96
408	Pregnancy outcome in patients with raised blood glucose due to a heterozygous glucokinase gene mutation. *Diabetic Medicine*, **2009**, 26, 14-8	3.5	94
407	Mutations in the genes encoding the pancreatic beta-cell KATP channel subunits Kir6.2 (KCNJ11) and SUR1 (ABCC8) in diabetes mellitus and hyperinsulinism. *Human Mutation*, **2006**, 27, 220-31	4.7	93
406	HLA genotyping supports a nonautoimmune etiology in patients diagnosed with diabetes under the age of 6 months. *Diabetes*, **2006**, 55, 1895-8	0.9	92
405	The 0.1% of the population with glucokinase monogenic diabetes can be recognized by clinical characteristics in pregnancy: the Atlantic Diabetes in Pregnancy cohort. *Diabetes Care*, **2014**, 37, 1230-6	14.6	90
404	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotype-phenotype correlation in an international cohort of patients. *Diabetologia*, **2013**, 56, 758-62	10.3	90
403	Beta-cell dysfunction, insulin sensitivity, and glycosuria precede diabetes in hepatocyte nuclear factor-1alpha mutation carriers. *Diabetes Care*, **2005**, 28, 1751-6	14.6	89
402	Clinical heterogeneity in patients with FOXP3 mutations presenting with permanent neonatal diabetes. *Diabetes Care*, **2009**, 32, 111-6	14.6	86

401	Activating glucokinase (GCK) mutations as a cause of medically responsive congenital hyperinsulinism: prevalence in children and characterisation of a novel GCK mutation. *European Journal of Endocrinology*, **2008**, 159, 27-34	6.5	84
400	Intrauterine hyperglycemia is associated with an earlier diagnosis of diabetes in HNF-1alpha gene mutation carriers. *Diabetes Care*, **2002**, 25, 2287-91	14.6	84
399	Hepatocyte nuclear factor-1beta: a new kindred with renal cysts and diabetes and gene expression in normal human development. *Journal of the American Society of Nephrology: JASN*, **2001**, 12, 2175-2180	12.7	84
398	Use of HbA1c in the identification of patients with hyperglycaemia caused by a glucokinase mutation: observational case control studies. *PLoS ONE*, **2013**, 8, e65326	3.7	83
397	SLC2A2 mutations can cause neonatal diabetes, suggesting GLUT2 may have a role in human insulin secretion. *Diabetologia*, **2012**, 55, 2381-5	10.3	82
396	Extreme phenotypic diversity and nonpenetrance in families with the LMNA gene mutation R644C. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1530-42	2.5	82
395	Prevalence of permanent neonatal diabetes in Slovakia and successful replacement of insulin with sulfonylurea therapy in KCNJ11 and ABCC8 mutation carriers. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 1276-82	5.6	82
394	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. *Journal of Clinical Endocrinology and Metabolism*, **2004**, 89, 3932-5	5.6	82
393	GATA4 mutations are a cause of neonatal and childhood-onset diabetes. *Diabetes*, **2014**, 63, 2888-94	0.9	80
392	Hepatocyte nuclear factor-1beta gene deletions--a common cause of renal disease. *Nephrology Dialysis Transplantation*, **2008**, 23, 627-35	4.3	79
391	Detection of an MEN1 gene mutation depends on clinical features and supports current referral criteria for diagnostic molecular genetic testing. *Clinical Endocrinology*, **2005**, 62, 169-75	3.4	79
390	Sirolimus therapy following subtotal pancreatectomy in neonatal hyperinsulinemic hypoglycaemia: a case report. *International Journal of Pediatric Endocrinology (Springer)*, **2015**, 2015,	1.5	78
389	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. *Acta Neuropathologica Communications*, **2016**, 4, 56	7.3	77
388	Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes due to KCNJ11 mutations: an international cohort study. *Lancet Diabetes and Endocrinology,the*, **2018**, 6, 637-646	18.1	77
387	Most people with long-duration type 1 diabetes in a large population-based study are insulin microsecretors. *Diabetes Care*, **2015**, 38, 323-8	14.6	76
386	High-sensitivity CRP discriminates HNF1A-MODY from other subtypes of diabetes. *Diabetes Care*, **2011**, 34, 1860-2	14.6	76
385	Increased all-cause and cardiovascular mortality in monogenic diabetes as a result of mutations in the HNF1A gene. *Diabetic Medicine*, **2010**, 27, 157-61	3.5	76
384	Insights into the structure and regulation of glucokinase from a novel mutation (V62M), which causes maturity-onset diabetes of the young. *Journal of Biological Chemistry*, **2005**, 280, 14105-13	5.4	76

383	tRNA methyltransferase homolog gene TRMT10A mutation in young onset diabetes and primary microcephaly in humans. *PLoS Genetics*, **2013**, 9, e1003888	6	75
382	Novel GLIS3 mutations demonstrate an extended multisystem phenotype. *European Journal of Endocrinology*, **2011**, 164, 437-43	6.5	75
381	High-dose glibenclamide can replace insulin therapy despite transitory diarrhea in early-onset diabetes caused by a novel R201L Kir6.2 mutation. *Diabetes Care*, **2005**, 28, 758-9	14.6	75
380	Concordance of assays designed for the quantification of JAK2V617F: a multicenter study. *Haematologica*, **2009**, 94, 38-45	6.6	74
379	Identifying hepatic nuclear factor 1alpha mutations in children and young adults with a clinical diagnosis of type 1 diabetes. *Diabetes Care*, **2003**, 26, 333-7	14.6	74
378	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. *Journal of the American Society of Nephrology: JASN*, **2017**, 28, 2529-2539	12.7	73
377	Population-Based Assessment of a Biomarker-Based Screening Pathway to Aid Diagnosis of Monogenic Diabetes in Young-Onset Patients. *Diabetes Care*, **2017**, 40, 1017-1025	14.6	73
376	Micronucleus assays using cytochalasin-blocked MCL-5 cells, a proprietary human cell line expressing five human cytochromes P-450 and microsomal epoxide hydrolase. *Mutagenesis*, **1993**, 8, 363-72	2.8	71
375	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. *Diabetologia*, **2005**, 48, 1029-31	10.3	69
374	The mutated human gene encoding hepatocyte nuclear factor 1beta inhibits kidney formation in developing Xenopus embryos. *Proceedings of the National Academy of Sciences of the United States of America*, **2000**, 97, 4695-700	11.5	69
373	Distinct molecular and morphogenetic properties of mutations in the human HNF1beta gene that lead to defective kidney development. *Journal of the American Society of Nephrology: JASN*, **2003**, 14, 2033-41	12.7	68
372	Diabetes mellitus in neonates and infants: genetic heterogeneity, clinical approach to diagnosis, and therapeutic options. *Hormone Research in Paediatrics*, **2013**, 80, 137-46	3.3	67
371	Next-generation sequencing reveals deep intronic cryptic ABCC8 and HADH splicing founder mutations causing hyperinsulinism by pseudoexon activation. *American Journal of Human Genetics*, **2013**, 92, 131-6	11	66
370	The use of genetically engineered V79 Chinese hamster cultures expressing rat liver CYP1A1, 1A2 and 2B1 cDNAs in micronucleus assays. *Mutagenesis*, **1991**, 6, 461-70	2.8	66
369	Hyperinsulinism-hyperammonaemia syndrome: novel mutations in the GLUD1 gene and genotype-phenotype correlations. *European Journal of Endocrinology*, **2009**, 161, 731-5	6.5	65
368	A gene for autosomal recessive spondylocostal dysostosis maps to 19q13.1-q13.3. *American Journal of Human Genetics*, **1999**, 65, 175-82	11	65
367	3-Hydroxyacyl-coenzyme A dehydrogenase deficiency and hyperinsulinemic hypoglycemia: characterization of a novel mutation and severe dietary protein sensitivity. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 2221-5	5.6	64
366	Mutations in the MESP2 gene cause spondylothoracic dysostosis/Jarcho-Levin syndrome. *American Journal of Human Genetics*, **2008**, 82, 1334-41	11	64

365	Entities and frequency of neonatal diabetes: data from the diabetes documentation and quality management system (DPV). *Diabetic Medicine*, **2010**, 27, 709-12	3.5	63
364	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. *American Journal of Human Genetics*, **2018**, 102, 27-43	11	61
363	Incidence, genetics, and clinical phenotype of permanent neonatal diabetes mellitus in northwest Saudi Arabia. *Pediatric Diabetes*, **2012**, 13, 499-505	3.6	60
362	Permanent neonatal diabetes due to activating mutations in ABCC8 and KCNJ11. *Reviews in Endocrine and Metabolic Disorders*, **2010**, 11, 193-8	10.5	60
361	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
360	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. *Nature Communications*, **2017**, 8, 888	17.4	57
359	The HNF4A R76W mutation causes atypical dominant Fanconi syndrome in addition to a □cell phenotype. *Journal of Medical Genetics*, **2014**, 51, 165-9	5.8	57
358	Dominant ER Stress-Inducing Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. *Diabetes*, **2017**, 66, 2044-2053	0.9	56
357	Mutations at the same residue (R50) of Kir6.2 (KCNJ11) that cause neonatal diabetes produce different functional effects. *Diabetes*, **2006**, 55, 1705-12	0.9	56
356	A missense mutation in the hepatocyte nuclear factor 4 alpha gene in a UK pedigree with maturity-onset diabetes of the young. *Diabetologia*, **1997**, 40, 859-62	10.3	55
355	The diabetic phenotype in HNF4A mutation carriers is moderated by the expression of HNF4A isoforms from the P1 promoter during fetal development. *Diabetes*, **2008**, 57, 1745-52	0.9	55
354	A genome-wide scan in families with maturity-onset diabetes of the young: evidence for further genetic heterogeneity. *Diabetes*, **2003**, 52, 872-81	0.9	55
353	Partial and whole gene deletion mutations of the GCK and HNF1A genes in maturity-onset diabetes of the young. *Diabetologia*, **2007**, 50, 2313-7	10.3	54
352	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. *Diabetologia*, **2016**, 59, 1162-6	10.3	54
351	Expanding the Clinical Spectrum Associated With GLIS3 Mutations. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, E1362-9	5.6	53
350	C282Y mutation in HFE (haemochromatosis) gene and type 2 diabetes. *Lancet, The*, **1998**, 351, 1933-4	40	53
349	Response to treatment with rosiglitazone in familial partial lipodystrophy due to a mutation in the LMNA gene. *Diabetic Medicine*, **2003**, 20, 823-7	3.5	53
348	Cellular and chromosomal hypersensitivity to DNA crosslinking agents and topoisomerase inhibitors in the radiosensitive Chinese hamster irs mutants: phenotypic similarities to ataxia telangiectasia and Fanconi anaemia cells. *Carcinogenesis*, **1993**, 14, 2487-94	4.6	52

347	Pitfalls of haplotype phasing from amplicon-based long-read sequencing. *Scientific Reports*, **2016**, 6, 21746	49	52
346	Pancreatic endocrine and exocrine function in children following near-total pancreatectomy for diffuse congenital hyperinsulinism. *PLoS ONE*, **2014**, 9, e98054	3.7	50
345	Etiological investigation of diabetes in young adults presenting with apparent type 2 diabetes. *Diabetes Care*, **2003**, 26, 2088-93	14.6	50
344	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. *Diabetes*, **2001**, 50, 2047-52	0.9	50
343	Confirmation of linkage of Duane's syndrome and refinement of the disease locus to an 8.8-cM interval on chromosome 2q31. *Human Genetics*, **2000**, 106, 636-8	6.3	50
342	KATP channel mutations in infants with permanent diabetes diagnosed after 6 months of life. *Pediatric Diabetes*, **2012**, 13, 322-5	3.6	49
341	An ABCC8 gene mutation and mosaic uniparental isodisomy resulting in atypical diffuse congenital hyperinsulinism. *Diabetes*, **2008**, 57, 259-63	0.9	49
340	Predictive genetic testing in maturity-onset diabetes of the young (MODY). *Diabetic Medicine*, **2001**, 18, 417-21	3.5	49
339	Permanent neonatal diabetes in an Asian infant. *Journal of Pediatrics*, **2005**, 146, 131-3	3.6	48
338	Diagnosis of lethal or prenatal-onset autosomal recessive disorders by parental exome sequencing. *Prenatal Diagnosis*, **2018**, 38, 33-43	3.2	47
337	Growth in PHEX-associated X-linked hypophosphatemic rickets: the importance of early treatment. *Pediatric Nephrology*, **2012**, 27, 581-8	3.2	47
336	Childhood presentation of COL4A1 mutations. *Developmental Medicine and Child Neurology*, **2012**, 54, 569-74	3.3	47
335	Increased ATPase activity produced by mutations at arginine-1380 in nucleotide-binding domain 2 of ABCC8 causes neonatal diabetes. *Proceedings of the National Academy of Sciences of the United States of America*, **2007**, 104, 18988-92	11.5	47
334	Frequent occurrence of an intron 4 mutation in multiple endocrine neoplasia type 1. *Journal of Clinical Endocrinology and Metabolism*, **2002**, 87, 2688-93	5.6	47
333	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. *European Journal of Human Genetics*, **2015**, 23, 401-4	5.3	46
332	Origin of de novo KCNJ11 mutations and risk of neonatal diabetes for subsequent siblings. *Journal of Clinical Endocrinology and Metabolism*, **2007**, 92, 1773-7	5.6	46
331	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. *Diabetes*, **2004**, 53, 2998-3001	0.9	46
330	Mutations in hepatocyte nuclear factor 1beta are not a common cause of maturity-onset diabetes of the young in the U.K. *Diabetes*, **1998**, 47, 1152-4	0.9	46

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