Sian Ellard

List of Publications by Year in Descending Order

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	Evaluation of Evidence for Pathogenicity Demonstrates that BLK, KLF11 and PAX4 Should not be Included in Diagnostic Testing for MODY.. *Diabetes*, **2022**,	0.9	2
489	SavvyCNV: Genome-wide CNV calling from off-target reads.. *PLoS Computational Biology*, **2022**, 18, e1009940	3	1
488	Congenital beta cell defects are not associated with markers of islet autoimmunity, even in the context of high genetic risk for type 1 diabetes.. *Diabetologia*, **2022**, 1	10.3	0
487	Refinements and considerations for trio whole-genome sequence analysis when investigating Mendelian diseases presenting in early childhood.. *Human Genetics and Genomics Advances*, **2022**, 3, 100113	0.8	0
486	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. *Diabetologia*, **2021**, 1	10.3	1
485	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
484	Syndromic Monogenic Diabetes Genes Should be Tested in Patients With a Clinical Suspicion of MODY. *Diabetes*, **2021**,	0.9	2
483	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). *Molecular Genetics & Genomic Medicine*, **2021**, e1674	2.3	0
482	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. *Genome Medicine*, **2021**, 13, 55	14.4	5
481	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. *Cell Reports*, **2021**, 35, 108981	10.6	4
480	Mutations in HID1 Cause Syndromic Infantile Encephalopathy and Hypopituitarism. *Annals of Neurology*, **2021**, 90, 143-158	9.4	1
479	Combining evidence for and against pathogenicity for variants in cancer susceptibility genes: CanVIG-UK consensus recommendations. *Journal of Medical Genetics*, **2021**, 58, 297-304	5.8	7
478	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. *Human Genetics and Genomics Advances*, **2021**, 2, 100015	0.8	2
477	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated Permanent Neonatal Diabetes. *Diabetes Care*, **2021**, 44, 35-42	14.6	7
476	Diagnostic RET genetic testing in 1,058 index patients: A UK centre perspective. *Clinical Endocrinology*, **2021**, 95, 295-302	3.4	0
475	Genotype and Phenotype Heterogeneity in Neonatal Diabetes: A Single Centre Experience in Turkey. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2021**, 13, 80-87	1.9	1
474	A hemizygous mutation in the FOXP3 gene (IPEX syndrome) resulting in recurrent X-linked fetal hydrops: a case report. *BMC Medical Genomics*, **2021**, 14, 58	3.7	1

473	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. *American Journal of Human Genetics*, **2021**, 108, 1342-1349	11	1
472	Molecular Genetics, Clinical Characteristics, and Treatment Outcomes of K-Channel Neonatal Diabetes Mellitus in Vietnam National Children's Hospital. *Frontiers in Endocrinology*, **2021**, 12, 727083	5.7	3
471	Study of Acute Liver Failure in Children Using Next Generation Sequencing Technology. *Journal of Pediatrics*, **2021**, 236, 124-130	3.6	1
470	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. *American Journal of Human Genetics*, **2020**, 107, 670-682	11	9
469	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. *Journal of Medical Genetics*, **2020**, 57, 829-834	5.8	11
468	Noninvasive Fetal Genotyping by Droplet Digital PCR to Identify Maternally Inherited Monogenic Diabetes Variants. *Clinical Chemistry*, **2020**, 66, 958-965	5.5	13
467	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389. *PLoS ONE*, **2020**, 15, e0228417	3.7	11
466	Update of variants identified in the pancreatic β-cell K channel genes KCNJ11 and ABCC8 in individuals with congenital hyperinsulinism and diabetes. *Human Mutation*, **2020**, 41, 884-905	4.7	37
465	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. *Genetics in Medicine*, **2020**, 22, 1005-1014	8.1	35
464	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. *Diabetes Care*, **2020**, 43, 82-89	14.6	35
463	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome. *Genetics in Medicine*, **2020**, 22, 867-877	8.1	17
462	Recurrent De Novo NAHR Reciprocal Duplications in the ATAD3 Gene Cluster Cause a Neurogenetic Trait with Perturbed Cholesterol and Mitochondrial Metabolism. *American Journal of Human Genetics*, **2020**, 106, 272-279	11	20
461	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. *Journal of Clinical Investigation*, **2020**, 130, 14-16	15.9	13
460	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. *Journal of Clinical Investigation*, **2020**, 130, 6338-6353	15.9	21
459	Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. *Journal of Clinical Endocrinology and Metabolism*, **2020**, 105,	5.6	21
458	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. *Wellcome Open Research*, **2020**, 5, 15	4.8	
457	Homozygous Hypomorphic Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. *Diabetes Care*, **2020**, 43, 909-912	14.6	7
456	Recurrent TTN metatranscript-only c.39974-11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. *Human Mutation*, **2020**, 41, 403-411	4.7	10

455	A novel autosomal recessive DEAF1 nonsense variant: expanding the clinical phenotype. *Clinical Dysmorphology*, **2020**, 29, 114-117	0.9	1
454	De Novo Mutations in Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. *Diabetes*, **2020**, 69, 477-483	0.9	17
453	Type 1 diabetes can present before the age of 6 months and is characterised by autoimmunity and rapid loss of beta cells. *Diabetologia*, **2020**, 63, 2605-2615	10.3	9
452	Response to Comment on Misra et al. Homozygous Hypomorphic Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care 2020;43:909-912. *Diabetes Care*, **2020**, 43, e155-e156	14.6	
451	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. *Wellcome Open Research*, **2020**, 5, 15	4.8	0
450	Compound heterozygous Pkd1l1 variants in a family with two fetuses affected by heterotaxy and complex Chd. *European Journal of Medical Genetics*, **2020**, 63, 103657	2.6	4
449	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389 **2020**, 15, e0228417		
448	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389 **2020**, 15, e0228417		
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446	Using referral rates for genetic testing to determine the incidence of a rare disease: The minimal incidence of congenital hyperinsulinism in the UK is 1 in 28,389 **2020**, 15, e0228417		
445	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. *Human Molecular Genetics*, **2019**, 28, 3543-3551	5.6	4
444	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. *Kidney International Reports*, **2019**, 4, 1304-1311	4.1	22
443	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. *American Journal of Human Genetics*, **2019**, 104, 985-989	11	28
442	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. *Diabetes*, **2019**, 68, 1528-1535	0.9	15
441	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 9 affected individuals. *Genetics in Medicine*, **2019**, 21, 233-242	8.1	23
440	The role of molecular genetics in the clinical management of sporadic medullary thyroid carcinoma: A systematic review. *Clinical Endocrinology*, **2019**, 91, 697-707	3.4	16
439	Genomic variant sharing: a position statement. *Wellcome Open Research*, **2019**, 4, 22	4.8	16
438	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. *Endocrinology, Diabetes and Metabolism Case Reports*, **2019**, 2019,	1.4	2

437	Congenital Hyperinsulinism and Evolution to Sulfonylurearesponsive Diabetes Later in Life due to a Novel Homozygous p.L171F Mutation. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2019**, 11, 82-87	1.9	10
436	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
435	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
434	Misannotation of multiple-nucleotide variants risks misdiagnosis. *Wellcome Open Research*, **2019**, 4, 145	4.8	0
433	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. *American Journal of Human Genetics*, **2019**, 105, 1286-1293	11	8
432	Using Structural Analysis to Assess the Impact of Missense Variants in MEN1. *Journal of the Endocrine Society*, **2019**, 3, 2258-2275	0.4	6
431	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
430	An Amish founder variant consolidates disruption of CEP55 as a cause of hydranencephaly and renal dysplasia. *European Journal of Human Genetics*, **2019**, 27, 657-662	5.3	17
429	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. *Genetics in Medicine*, **2019**, 21, 982-986	8.1	15
428	Focal Congenital Hyperinsulinism as a Cause for Sudden Infant Death. *Pediatric and Developmental Pathology*, **2019**, 22, 65-69	2.2	4
427	Risk category system to identify pituitary adenoma patients with mutations. *Journal of Medical Genetics*, **2018**, 55, 254-260	5.8	22
426	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. *Annals of Neurology*, **2018**, 83, 1105-1124	9.4	59
425	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. *Diabetologia*, **2018**, 61, 1027-1036	10.3	17
424	missense mutation causes familial insulinomatosis and diabetes mellitus. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, 1027-1032	11.5	45
423	A type 1 diabetes genetic risk score can discriminate monogenic autoimmunity with diabetes from early-onset clustering of polygenic autoimmunity with diabetes. *Diabetologia*, **2018**, 61, 862-869	10.3	20
422	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. *Diabetes Care*, **2018**, 41, e7	14.6	3
421	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. *American Journal of Human Genetics*, **2018**, 102, 27-43	11	61
420	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 195-204	1.6	8

419	Emergence of insulin resistance following empirical glibenclamide therapy: a case report of neonatal diabetes with a recessive INS gene mutation. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 345-348	1.6	7
418	Prevalence of diabetes in Australia: insights from the Fremantle Diabetes Study Phase II. *Internal Medicine Journal*, **2018**, 48, 803-809	1.6	19
417	Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation in NEUROD1. *Pediatric Diabetes*, **2018**, 19, 898-904	3.6	14
416	Diagnosis of lethal or prenatal-onset autosomal recessive disorders by parental exome sequencing. *Prenatal Diagnosis*, **2018**, 38, 33-43	3.2	47
415	Diazoxide toxicity in a child with persistent hyperinsulinemic hypoglycemia of infancy: mixed hyperglycemic hyperosmolar coma and ketoacidosis. *Journal of Pediatric Endocrinology and Metabolism*, **2018**, 31, 943-945	1.6	6
414	TRPV6 compound heterozygous variants result in impaired placental calcium transport and severe undermineralization and dysplasia of the fetal skeleton. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1950-1955	2.5	24
413	Comprehensive screening shows that mutations in the known syndromic genes are rare in infants presenting with hyperinsulinaemic hypoglycaemia. *Clinical Endocrinology*, **2018**, 89, 621-627	3.4	4
412	Exocrine pancreatic dysfunction is common in hepatocyte nuclear factor 1□associated renal disease and can be symptomatic. *CKJ: Clinical Kidney Journal*, **2018**, 11, 453-458	4.5	5
411	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
410	The Common Variant I27L Is a Modifier of Age at Diabetes Diagnosis in Individuals With HNF1A-MODY. *Diabetes*, **2018**, 67, 1903-1907	0.9	7
409	Neonatal Diabetes: Two Cases with Isolated Pancreas Agenesis due to Homozygous PTF1A Enhancer Mutations and One with Developmental Delay, Epilepsy, and Neonatal Diabetes Syndrome due to KCNJ11 Mutation. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2018**, 10, 168-174	1.9	12
408	Sirolimus-Induced Hepatitis in Two Patients with Hyperinsulinemic Hypoglycemia. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2018**, 10, 279-283	1.9	5
407	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling performance using the ICR142 NGS validation series. *Wellcome Open Research*, **2018**, 3, 108	4.8	
406	Monogenic Diabetes Not Caused By Mutations in Mody Genes: A Very Heterogenous Group of Diabetes. *Experimental and Clinical Endocrinology and Diabetes*, **2018**, 126, 612-618	2.3	5
405	Marked intrafamilial variability of exocrine and endocrine pancreatic phenotypes due to a splice site mutation in GATA6. *Biotechnology and Biotechnological Equipment*, **2018**, 32, 124-129	1.6	
404	A Novel KCNJ11 Mutation Associated with Transient Neonatal Diabetes. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2018**, 10, 175-178	1.9	6
403	p.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancer. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 4275-4282	5.6	24
402	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling using the ICR142 NGS validation series. *Wellcome Open Research*, **2018**, 3, 108	4.8	

401	Clinical Diversity in Focal Congenital Hyperinsulinism in Infancy Correlates With Histological Heterogeneity of Islet Cell Lesions. *Frontiers in Endocrinology*, **2018**, 9, 619	5.7	9
400	Response to Letter to the Editor: "p.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancer". *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 3518-3519	5.6	3
399	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. *Journal of Clinical Endocrinology and Metabolism*, **2018**, 103, 3225-3230	5.6	14
398	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
397	A UK nationwide prospective study of treatment change in MODY: genetic subtype and clinical characteristics predict optimal glycaemic control after discontinuing insulin and metformin. *Diabetologia*, **2018**, 61, 2520-2527	10.3	38
396	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
395	Recessively Inherited Mutations Cause Autoimmunity Presenting as Neonatal Diabetes. *Diabetes*, **2017**, 66, 2316-2322	0.9	39
394	A successful transition to sulfonylurea treatment in male infant with neonatal diabetes caused by the novel abcc8 gene mutation and three years follow-up. *Diabetes Research and Clinical Practice*, **2017**, 129, 59-61	7.4	5
393	Atypical Forms of Congenital Hyperinsulinism in Infancy Are Associated With Mosaic Patterns of Immature Islet Cells. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 3261-3267	5.6	21
392	Clinical presentation and treatment response to diazoxide in two siblings with congenital hyperinsulinism as a result of a novel compound heterozygous ABCC8 missense mutation. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 471-474	1.6	1
391	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
390	Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL). *Metabolism: Clinical and Experimental*, **2017**, 71, 213-225	12.7	31
389	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. *Nature Communications*, **2017**, 8, 888	17.4	57
388	The prevalence of monogenic diabetes in Australia: the Fremantle Diabetes Study Phase II. *Medical Journal of Australia*, **2017**, 207, 344-347	4	11
387	Fainting Fanconi syndrome clarified by proxy: a case report. *BMC Nephrology*, **2017**, 18, 230	2.7	8
386	MODY in Ukraine: genes, clinical phenotypes and treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 1095-1103	1.6	5
385	Analysis of large-scale sequencing cohorts does not support the role of variants in UCP2 as a cause of hyperinsulinaemic hypoglycaemia. *Human Mutation*, **2017**, 38, 1442-1444	4.7	15
384	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. *Diabetologia*, **2017**, 60, 2168-2173	10.3	7

383	In-frame seven amino-acid duplication in arose over the last 3000 years, disrupts protein interaction and stability and is associated with gigantism. *European Journal of Endocrinology*, **2017**, 177, 257-266	6.5	11
382	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
381	Clinical and Molecular Characterization of Children with Neonatal Diabetes Mellitus at a Tertiary Care Center in Northern India. *Indian Pediatrics*, **2017**, 54, 467-471	1.2	9
380	Case report: maternal mosaicism resulting in inheritance of a novel GATA6 mutation causing pancreatic agenesis and neonatal diabetes mellitus. *Diagnostic Pathology*, **2017**, 12, 1	3	20
379	Analysis of cell-free fetal DNA for non-invasive prenatal diagnosis in a family with neonatal diabetes. *Diabetic Medicine*, **2017**, 34, 582-585	3.5	24
378	Increased Population Risk of AIP-Related Acromegaly and Gigantism in Ireland. *Human Mutation*, **2017**, 38, 78-85	4.7	24
377	The Clinical Course of Patients with Preschool Manifestation of Type 1 Diabetes Is Independent of the HLA DR-DQ Genotype. *Genes*, **2017**, 8,	4.2	7
376	Pancreatic Agenesis due to Compound Heterozygosity for a Novel Enhancer and Truncating Mutation in the PTF1A Gene. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2017**, 9, 274-277	1.9	16
375	An Nonsense Mutation Causing Neonatal Diabetes Through Altered Transcript Expression. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2017**, 9, 260-264	1.9	10
374	Prematurity and Genetic Testing for Neonatal Diabetes. *Pediatrics*, **2016**, 138,	7.4	19
373	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
372	Prospective functional classification of all possible missense variants in PPARG. *Nature Genetics*, **2016**, 48, 1570-1575	36.3	149
371	Hyperinsulinemic Hypoglycemia of Infancy due to Novel HADH Mutation in Two Siblings. *Indian Pediatrics*, **2016**, 53, 912-913	1.2	4
370	Conservatively treated Congenital Hyperinsulinism (CHI) due to K-ATP channel gene mutations: reducing severity over time. *Orphanet Journal of Rare Diseases*, **2016**, 11, 163	4.2	32
369	Coexistence of Mosaic Uniparental Isodisomy and a KCNJ11 Mutation Presenting as Diffuse Congenital Hyperinsulinism and Hemihypertrophy. *Hormone Research in Paediatrics*, **2016**, 85, 421-5	3.3	6
368	Genetic characteristics, clinical spectrum, and incidence of neonatal diabetes in the Emirate of AbuDhabi, United Arab Emirates. *American Journal of Medical Genetics, Part A*, **2016**, 170, 602-9	2.5	26
367	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
366	Insights from Monogenic Diabetes **2016**, 223-240		

365	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. *Diabetes*, **2016**, 65, 2810-5	0.9	16
364	Somatic GPR101 Duplication Causing X-Linked Acrogigantism (XLAG)-Diagnosis and Management. *Journal of Clinical Endocrinology and Metabolism*, **2016**, 101, 1927-30	5.6	34
363	Characteristics of maturity onset diabetes of the young in a large diabetes center. *Pediatric Diabetes*, **2016**, 17, 360-7	3.6	36
362	Single patient in GCK-MODY family successfully re-diagnosed into GCK-PNDM through targeted next-generation sequencing technology. *Acta Diabetologica*, **2016**, 53, 337-8	3.9	2
361	A Novel Homozygous Mutation in the KCNJ11 Gene of a Neonate with Congenital Hyperinsulinism and Successful Management with Sirolimus. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2016**, 8, 478-481	1.9	15
360	Clinical and Genetic Characteristics, Management and Long-Term Follow-Up of Turkish Patients with Congenital Hyperinsulinism. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2016**, 8, 197-204	1.9	2
359	GCK gene mutations are a common cause of childhood-onset MODY (maturity-onset diabetes of the young) in Turkey. *Clinical Endocrinology*, **2016**, 85, 393-9	3.4	12
358	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. *Kidney International*, **2016**, 90, 203-11	9.9	42
357	SOS1 frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. *Clinical Endocrinology*, **2016**, 84, 715-9	3.4	7
356	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. *Diabetologia*, **2016**, 59, 2262-5	10.3	13
355	Pitfalls of haplotype phasing from amplicon-based long-read sequencing. *Scientific Reports*, **2016**, 6, 21746	4.9	52
354	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
353	AIP mutations in young patients with acromegaly and the Tampico Giant: the Mexican experience. *Endocrine*, **2016**, 53, 402-11	4	14
352	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. *Diabetes*, **2016**, 65, 2094-2099	0.9	105
351	Differential regulation of serum microRNA expression by HNF1α and HNF1β transcription factors. *Diabetologia*, **2016**, 59, 1463-1473	10.3	15
350	Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in ATP sensitivity markedly affect diabetes risk. *Diabetologia*, **2016**, 59, 1430-1436	10.3	18
349	Clinical and genetic features of Argentinian children with diabetes-onset before 12months of age: Successful transfer from insulin to oral sulfonylurea. *Diabetes Research and Clinical Practice*, **2016**, 117, 104-10	7.4	7
348	The Common p.R114W HNF4A Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. *Diabetes*, **2016**, 65, 3212-7	0.9	29

347	Diagnosis of monogenic diabetes: 10-Year experience in a large multi-ethnic diabetes center. *Journal of Diabetes Investigation*, **2016**, 7, 332-7	3.9	14
346	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. *European Journal of Human Genetics*, **2015**, 23, 401-4	5.3	46
345	Efficacy and safety of sirolimus in a neonate with persistent hypoglycaemia following near-total pancreatectomy for hyperinsulinaemic hypoglycaemia. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1391-8	1.6	15
344	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
343	Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation. *Diabetes Care*, **2015**, 38, 1383-92	14.6	157
342	A Deep Intronic HADH Splicing Mutation (c.636+471G>T) in a Congenital Hyperinsulinemic Hypoglycemia Case: Long Term Clinical Course. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2015**, 7, 144-7	1.9	10
341	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
340	Variable Phenotype of Diabetes Mellitus in Siblings with a Homozygous PTF1A Enhancer Mutation. *Hormone Research in Paediatrics*, **2015**, 84, 206-11	3.3	10
339	Liver disease and other comorbidities in Wolcott-Rallison syndrome: different phenotype and variable associations in a large cohort. *Hormone Research in Paediatrics*, **2015**, 83, 190-7	3.3	19
338	Sirolimus therapy in a patient with severe hyperinsulinaemic hypoglycaemia due to a compound heterozygous ABCC8 gene mutation. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 695-9	1.6	19
337	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. *European Journal of Endocrinology*, **2015**, 172, 697-705	6.5	40
336	Genotype and phenotype correlations in Iranian patients with hyperinsulinaemic hypoglycaemia. *BMC Research Notes*, **2015**, 8, 350	2.3	6
335	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
334	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. *European Journal of Human Genetics*, **2015**, 23, 1744-8	5.3	24
333	Protein-induced hyperinsulinaemic hypoglycaemia due to a homozygous HADH mutation in three siblings of a Saudi family. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1073-7	1.6	15
332	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. *Journal of Pediatric Endocrinology and Metabolism*, **2015**, 28, 1279-86	1.6	25
331	HNF1B-associated renal and extra-renal disease-an expanding clinical spectrum. *Nature Reviews Nephrology*, **2015**, 11, 102-12	14.9	163
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5	Congenital hyperinsulinism and evolution to sulfonylurea-responsive diabetes later in life due to a novel homozygous p.L171F ABCC8 mutation. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*,	1.9	2
4	Using structural analysis in silico to assess the impact of missense variants in MEN1		1
3	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes		1
2	SavvyCNV: genome-wide CNV calling from off-target reads		8
1	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. *Wellcome Open Research*,2, 49	4.8	1