Sian Ellard

List of Publications by Year
in descending order

497 papers	38,456 citations	2423 97 h-index	3997 176 g-index
519 all docs	519 docs citations	519 times ranked	28293 citing authors

#	Article	IF	Citations
1	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. Diabetologia, 2022, 65, 336-342.	2.9	12
2	Quantifying prediction of pathogenicity for within-codon concordance (PM5) using 7541 functional classifications of BRCA1 and MSH2 missense variants. Genetics in Medicine, 2022, 24, 552-563.	1.1	5
3	Syndromic Monogenic Diabetes Genes Should Be Tested in Patients With a Clinical Suspicion of Maturity-Onset Diabetes of the Young. Diabetes, 2022, 71, 530-537.	0.3	35
4	Improvements in Awareness and Testing Have Led to a Threefold Increase Over 10 Years in the Identification of Monogenic Diabetes in the U.K.. Diabetes Care, 2022, 45, 642-649.	4.3	17
5	Evaluation of Evidence for Pathogenicity Demonstrates That BLK, KLF11, and PAX4 Should Not Be Included in Diagnostic Testing for MODY. Diabetes, 2022, 71, 1128-1136.	0.3	27
6	SavvyCNV: Genome-wide CNV calling from off-targetÂreads. PLoS Computational Biology, 2022, 18, e1009940.	1.5	18
7	THUMPD1 bi-allelic variants cause loss of tRNA acetylation and a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 587-600.	2.6	19
8	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.	2.9	1
9	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.	1.0	4
10	Combining evidence for and against pathogenicity for variants in cancer susceptibility genes: CanVIG-UK consensus recommendations. Journal of Medical Genetics, 2021, 58, 297-304.	1.5	28
11	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.	1.0	6
12	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated ABCC8 Permanent Neonatal Diabetes. Diabetes Care, 2021, 44, 35-42.	4.3	24
13	Diagnostic RET genetic testing in 1,058 index patients: A UK centre perspective. Clinical Endocrinology, 2021, 95, 295-302.	1.2	3
14	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.	0.4	3
15	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.	0.7	1
16	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular Genetics & Genomic Medicine, 2021, , e1674.	0.6	2
17	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	3.6	16
18	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. Cell Reports, 2021, 35, 108981.	2.9	9

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19	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	2.8	3
20	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.	2.6	9
21	Molecular Genetics, Clinical Characteristics, and Treatment Outcomes of KATP-Channel Neonatal Diabetes Mellitus in Vietnam National Childrenâ€™s Hospital. Frontiers in Endocrinology, 2021, 12, 727083.	1.5	4
22	Study of Acute Liver Failure in Children Using Next Generation Sequencing Technology. Journal of Pediatrics, 2021, 236, 124-130.	0.9	7
23	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	13.9	352
24	Lissencephaly-pachygyria spectrum in a North Indian boy with Wolcott-Rallison syndrome due to homozygous deletion of exon 1 in the EIF2AK3 gene. Pediatric Endocrinology, Diabetes and Metabolism, 2021, 27, 287-290.	0.3	1
25	Compound heterozygous Pkd1l1 variants in a family with two fetuses affected by heterotaxy and complex Chd. European Journal of Medical Genetics, 2020, 63, 103657.	0.7	12
26	Recurrent TTN metatranscriptâ€only c.39974â€"11T>G splice variant associated with autosomal recessive arthrogryposis multiplex congenita and myopathy. Human Mutation, 2020, 41, 403-411.	1.1	28
27	A novel autosomal recessive DEAF1 nonsense variant: expanding the clinical phenotype. Clinical Dysmorphology, 2020, 29, 114-117.	0.1	1
28	De Novo Mutations in EIF2B1 Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. Diabetes, 2020, 69, 477-483.	0.3	29
29	Type 1 diabetes can present before the age of 6Âmonths and is characterised by autoimmunity and rapid loss of beta cells. Diabetologia, 2020, 63, 2605-2615.	2.9	24
30	Response to Comment on Misra et al. Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care 2020;43:909â€"912. Diabetes Care, 2020, 43, e155-e156.	4.3	0
31	Unsupervised Clustering of Missense Variants in HNF1A Using Multidimensional Functional Data Aids Clinical Interpretation. American Journal of Human Genetics, 2020, 107, 670-682.	2.6	25
32	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.	1.5	30
33	Noninvasive Fetal Genotyping by Droplet Digital PCR to Identify Maternally Inherited Monogenic Diabetes Variants. Clinical Chemistry, 2020, 66, 958-965.	1.5	32
34	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.	1.1	29
35	Update of variants identified in the pancreatic Î²â€cell K _{ATP} channel genes KCNJ11 and ABCC8 in individuals with congenital hyperinsulinism and diabetes. Human Mutation, 2020, 41, 884-905.	1.1	90
36	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99

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37	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.	4.3	68
38	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	1.1	41
39	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.	2.6	33
40	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. Wellcome Open Research, 2020, 5, 15.	0.9	1
41	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, 2020, 130, 6338-6353.	3.9	58
42	Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors. Journal of Clinical Endocrinology and Metabolism, 2020, 105, e2247-e2260.	1.8	37
43	Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care, 2020, 43, 909-912.	4.3	13
44	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. Wellcome Open Research, 2020, 5, 15.	0.9	1
45	Title is missing!. , 2020, 15, e0228417.		0
46	Title is missing!. , 2020, 15, e0228417.		0
47	Title is missing!. , 2020, 15, e0228417.		0
48	Title is missing!. , 2020, 15, e0228417.		0
49	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 10 affected individuals. Genetics in Medicine, 2019, 21, 233-242.	1.1	39
50	The role of molecular genetics in the clinical management of sporadic medullary thyroid carcinoma: A systematic review. Clinical Endocrinology, 2019, 91, 697-707.	1.2	25
51	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	1.4	9
52	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.4	39
53	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.	2.6	43
54	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. Diabetes, 2019, 68, 1528-1535.	0.3	22

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55	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	2.6	18
56	Using Structural Analysis In Silico to Assess the Impact of Missense Variants in MEN1. Journal of the Endocrine Society, 2019, 3, 2258-2275.	0.1	14
57	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
58	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.	1.4	24
59	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. Genetics in Medicine, 2019, 21, 982-986.	1.1	22
60	Focal Congenital Hyperinsulinism as a Cause for Sudden Infant Death. Pediatric and Developmental Pathology, 2019, 22, 65-69.	0.5	5
61	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.	3.9	27
62	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	0.9	31
63	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, .	0.2	4
64	Congenital Hyperinsulinism and Evolution to Sulfonylurearesponsive Diabetes Later in Life due to a Novel Homozygous p.L171F ABCC8 Mutation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 82-87.	0.4	18
65	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	0.9	3
66	Refinement of the critical genomic region for congenital hyperinsulinismÂin the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	0.9	5
67	Misannotation of multiple-nucleotide variants risks misdiagnosis. Wellcome Open Research, 2019, 4, 145.	0.9	1
68	Risk category system to identify pituitary adenoma patients with AIP mutations. Journal of Medical Genetics, 2018, 55, 254-260.	1.5	35
69	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
70	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. Diabetologia, 2018, 61, 1027-1036.	2.9	26
71	MAFA 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.	3.3	88
72	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.	2.9	33

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73	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 (HNF1B) Molecular Defects. Diabetes Care 2017;40:1436â€“1443. Diabetes Care, 2018, 41, e7-e7.	4.3	4
74	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
75	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 195-204.	0.4	16
76	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.	0.4	8
77	Prevalence of diabetes in Australia: insights from the Fremantle Diabetes Study Phase II. Internal Medicine Journal, 2018, 48, 803-809.	0.5	46
78	Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation inNEUROD1. Pediatric Diabetes, 2018, 19, 898-904.	1.2	22
79	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	1.1	64
80	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.	0.6	12
81	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.	0.5	0
82	A Novel KCNJ11 Mutation Associated with Transient Neonatal Diabetes. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 175-178.	0.4	9
83	Cover Image, Volume 176A, Number 9, September 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
84	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.	1.8	39
85	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
86	Clinical Diversity in Focal Congenital Hyperinsulinism in Infancy Correlates With Histological Heterogeneity of Islet Cell Lesions. Frontiers in Endocrinology, 2018, 9, 619.	1.5	12
87	Response to Letter to the Editor: â€œp.Val804Met, the Most Frequent Pathogenic Mutation in RET, Confers a Very Low Lifetime Risk of Medullary Thyroid Cancerâ€• Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3518-3519.	1.8	3
88	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.	1.8	19
89	ISPAD Clinical Practice Consensus Guidelines 2018: The diagnosis and management of monogenic diabetes in children and adolescents. Pediatric Diabetes, 2018, 19, 47-63.	1.2	227
90	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.	2.9	65

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91	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.	0.4	7
92	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.	0.7	31
93	Comprehensive screening shows that mutations in the known syndromic genes are rare in infants presenting with hyperinsulinaemic hypoglycaemia. Clinical Endocrinology, 2018, 89, 621-627.	1.2	5
94	Exocrine pancreatic dysfunction is common in hepatocyte nuclear factor 1Î²-associated renal disease and can be symptomatic. CKJ: Clinical Kidney Journal, 2018, 11, 453-458.	1.4	10
95	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.	5.5	120
96	The Common HNF1A Variant I27L Is a Modifier of Age at Diabetes Diagnosis in Individuals With HNF1A-MODY. Diabetes, 2018, 67, 1903-1907.	0.3	12
97	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.	0.4	19
98	Sirolimus-Induced Hepatitis in Two Patients with Hyperinsulinemic Hypoglycemia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 279-283.	0.4	10
99	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling performance using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
100	Dominant ER Stressâ€“Inducing WFS1 Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. Diabetes, 2017, 66, 2044-2053.	0.3	77
101	Recessively InheritedLRBAMutations Cause Autoimmunity Presenting as Neonatal Diabetes. Diabetes, 2017, 66, 2316-2322.	0.3	59
102	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.	1.1	6
103	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.	1.8	24
104	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.	0.4	1
105	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.	3.0	99
106	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.	1.5	43
107	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. Nature Communications, 2017, 8, 888.	5.8	95
108	MODY in Ukraine: genes, clinical phenotypes and treatment. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1095-1103.	0.4	7

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109	Analysis of large-scale sequencing cohorts does not support the role of variants inUCP2as a cause of hyperinsulinaemic hypoglycaemia. Human Mutation, 2017, 38, 1442-1444.	1.1	17
110	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. Diabetologia, 2017, 60, 2168-2173.	2.9	12
111	In-frame seven amino-acid duplication in AIP arose over the last 3000 years, disrupts protein interaction and stability and is associated with gigantism. European Journal of Endocrinology, 2017, 177, 257-266.	1.9	12
112	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.	4.3	111
113	Clinical and molecular characterization of children with neonatal diabetes mellitus at a tertiary care center in northern India. Indian Pediatrics, 2017, 54, 467-471.	0.2	13
114	Case report: maternal mosaicism resulting in inheritance of a novel GATA6 mutation causing pancreatic agenesis and neonatal diabetes mellitus. Diagnostic Pathology, 2017, 12, 1.	0.9	33
115	Analysis of cellâ€free fetal <scp>DNA</scp> for nonâ€invasive prenatal diagnosis in a family with neonatal diabetes. Diabetic Medicine, 2017, 34, 582-585.	1.2	27
116	Increased Population Risk ofAIP-Related Acromegaly and Gigantism in Ireland. Human Mutation, 2017, 38, 78-85.	1.1	25
117	The Clinical Course of Patients with Preschool Manifestation of Type 1 Diabetes Is Independent of the HLA DR-DQ Genotype. Genes, 2017, 8, 146.	1.0	9
118	The prevalence of monogenic diabetes in Australia: the Fremantle Diabetes Study Phase II. Medical Journal of Australia, 2017, 207, 344-347.	0.8	18
119	Fainting Fanconi syndrome clarified by proxy: a case report. BMC Nephrology, 2017, 18, 230.	0.8	12
120	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.	0.4	23
121	An ABCC8 Nonsense Mutation Causing Neonatal Diabetes Through Altered Transcript Expression. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 260-264.	0.4	13
122	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.	0.4	4
123	<scp>GCK</scp> gene mutations are a common cause of childhoodâ€onset <scp>MODY</scp> (maturityâ€onset diabetes of the young) in Turkey. Clinical Endocrinology, 2016, 85, 393-399.	1.2	21
124	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. Kidney International, 2016, 90, 203-211.	2.6	64
125	<scp>SOS</scp>1 frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. Clinical Endocrinology, 2016, 84, 715-719.	1.2	11
126	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, 2262-2265.	2.9	28

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127	Pitfalls of haplotype phasing from amplicon-based long-read sequencing. Scientific Reports, 2016, 6, 21746.	1.6	62
128	Successful transfer to sulfonylureas in KCNJ11 neonatal diabetes is determined by the mutation and duration of diabetes. Diabetologia, 2016, 59, 1162-1166.	2.9	68
129	AIP mutations in young patients with acromegaly and the Tampico Giant: the Mexican experience. Endocrine, 2016, 53, 402-411.	1.1	20
130	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
131	Differential regulation of serum microRNA expression by HNF1Î² and HNF1Î± transcription factors. Diabetologia, 2016, 59, 1463-1473.	2.9	18
132	Neonatal diabetes caused by a homozygous KCNJ11 mutation demonstrates that tiny changes in ATP sensitivity markedly affect diabetes risk. Diabetologia, 2016, 59, 1430-1436.	2.9	25
133	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, 104-110.	1.1	8
134	The Common p.R114W HNF4A Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. Diabetes, 2016, 65, 3212-3217.	0.3	46
135	Diagnosis of monogenic diabetes: 10â€Year experience in a large multiâ€ethnic diabetes center. Journal of Diabetes Investigation, 2016, 7, 332-337.	1.1	21
136	Prematurity and Genetic Testing for Neonatal Diabetes. Pediatrics, 2016, 138, .	1.0	27
137	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56.	2.4	110
138	Prospective functional classification of all possible missense variants in PPARG. Nature Genetics, 2016, 48, 1570-1575.	9.4	210
139	Hyperinsulinemic hypoglycemia of infancy due to novel HADH mutation in two siblings. Indian Pediatrics, 2016, 53, 912-913.	0.2	6
140	Conservatively treated Congenital Hyperinsulinism (CHI) due to K-ATP channel gene mutations: reducing severity over time. Orphanet Journal of Rare Diseases, 2016, 11, 163.	1.2	42
141	Coexistence of Mosaic Uniparental Isodisomy and a <i>KCNJ11</i> Mutation Presenting as Diffuse Congenital Hyperinsulinism and Hemihypertrophy. Hormone Research in Paediatrics, 2016, 85, 421-425.	0.8	7
142	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, 602-609.	0.7	39
143	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.	4.3	172
144	Insights from Monogenic Diabetes. , 2016, , 223-240.		0

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145	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. Diabetes, 2016, 65, 2810-2815.	0.3	22
146	SomaticGPR101Duplication Causing X-Linked Acrogigantism (XLAG)â€"Diagnosis and Management. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1927-1930.	1.8	48
147	Characteristics of maturity onset diabetes of the young in a large diabetes center. Pediatric Diabetes, 2016, 17, 360-367.	1.2	44
148	Single patient in GCK-MODY family successfully re-diagnosed into GCK-PNDM through targeted next-generation sequencing technology. Acta Diabetologica, 2016, 53, 337-338.	1.2	3
149	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.	0.4	17
150	A Case Report on Congenital Hyperinsulinism Associated with ABCC8 Nonsense Mutation: Good Response to Octreotide. Journal of the ASEAN Federation of Endocrine Societies, 2016, 31, 178-182.	0.1	0
151	The value of inÂvitro studies in a case of neonatal diabetes with a novel Kir6.2â€W68G mutation. Clinical Case Reports (discontinued), 2015, 3, 884-887.	0.2	4
152	Sirolimus therapy following subtotal pancreatectomy in neonatal hyperinsulinemic hypoglycaemia: a case report. International Journal of Pediatric Endocrinology (Springer), 2015, 2015, .	1.6	0
153	Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 345-51.	0.4	10
154	SP030HNF1B WHOLE-GENE DELETIONS ARE ASSOCIATED WITH AUTISTIC TRAITS. Nephrology Dialysis Transplantation, 2015, 30, iii390-iii390.	0.4	0
155	Anemia in a Child with Deafness: Be Vigilant for a Rare Cause!. Indian Journal of Hematology and Blood Transfusion, 2015, 31, 394-395.	0.3	0
156	Most People With Long-Duration Type 1 Diabetes in a Large Population-Based Study Are Insulin Microsecretors. Diabetes Care, 2015, 38, 323-328.	4.3	104
157	Genome, Exome, and Targeted Next-Generation Sequencing in Neonatal Diabetes. Pediatric Clinics of North America, 2015, 62, 1037-1053.	0.9	16
158	Expanding the Clinical Spectrum Associated WithGLIS3Mutations. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1362-E1369.	1.8	66
159	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. Epilepsy Research, 2015, 118, 22-28.	0.8	6
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