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	A Common Variant in the FTO Gene Is Associated with Body Mass Index and Predisposes to Childhood and Adult Obesity. Science, 2007, 316, 889-894.	6.0	3,884
2	Replication of Genome-Wide Association Signals in UK Samples Reveals Risk Loci for Type 2 Diabetes. Science, 2007, 316, 1336-1341.	6.0	2,040
3	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, 1838-1849.	13.9	1,077
4	Switching from Insulin to Oral Sulfonylureas in Patients with Diabetes Due to Kir6.2 Mutations. New England Journal of Medicine, 2006, 355, 467-477.	13.9	878
5	Mutations in the glucokinase gene of the fetus result in reduced birth weight. Nature Genetics, 1998, 19, 268-270.	9.4	565
6	Maturity-onset diabetes of the young (MODY): how many cases are we missing?. Diabetologia, 2010, 53, 2504-2508.	2.9	560
7	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, 15040-15044.	3.3	494
8	Hypomethylation of multiple imprinted loci in individuals with transient neonatal diabetes is associated with mutations in ZFP57. Nature Genetics, 2008, 40, 949-951.	9.4	460
9	Clinical implications of a molecular genetic classification of monogenic Î²-cell diabetes. Nature Clinical Practice Endocrinology and Metabolism, 2008, 4, 200-213.	2.9	439
10	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. Nature Genetics, 2014, 46, 812-814.	9.4	411
11	Update on mutations in glucokinase (GCK), which cause maturity-onset diabetes of the young, permanent neonatal diabetes, and hyperinsulinemic hypoglycemia. Human Mutation, 2009, 30, 1512-1526.	1.1	403
12	Best practice guidelines for the molecular genetic diagnosis of maturity-onset diabetes of the young. Diabetologia, 2008, 51, 546-553.	2.9	376
13	Mutations in the human Delta homologue, DLL3, cause axial skeletal defects in spondylocostal dysostosis. Nature Genetics, 2000, 24, 438-441.	9.4	362
14	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
15	Macrosomia and Hyperinsulinaemic Hypoglycaemia in Patients with Heterozygous Mutations in the HNF4A Gene. PLoS Medicine, 2007, 4, e118.	3.9	349
16	Insulin Mutation Screening in 1,044 Patients With Diabetes. Diabetes, 2008, 57, 1034-1042.	0.3	347
17	Using SIFT and PolyPhen to Predict Loss-of-Function and Gain-of-Function Mutations. Genetic Testing and Molecular Biomarkers, 2010, 14, 533-537.	0.3	330
18	Mutations in ATP-Sensitive K+ Channel Genes Cause Transient Neonatal Diabetes and Permanent Diabetes in Childhood or Adulthood. Diabetes, 2007, 56, 1930-1937.	0.3	320

#	Article	IF	Citations
19	Alagille syndrome: pathogenesis, diagnosis and management. European Journal of Human Genetics, 2012, 20, 251-257.	1.4	319
20	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, 4055-4062.	1.8	310
21	Mutations in hepatocyte nuclear factor-1Â and their related phenotypes. Journal of Medical Genetics, 2005, 43, 84-90.	1.5	291
22	Mutations in the Hepatocyte Nuclear Factor-1Î² Gene Are Associated with Familial Hypoplastic Glomerulocystic Kidney Disease. American Journal of Human Genetics, 2001, 68, 219-224.	2.6	263
23	Prevalence of Vascular Complications Among Patients With Glucokinase Mutations and Prolonged, Mild Hyperglycemia. JAMA - Journal of the American Medical Association, 2014, 311, 279.	3.8	257
24	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	9.4	255
25	The effect of early, comprehensive genomic testing on clinical care in neonatal diabetes: an international cohort study. Lancet, The, 2015, 386, 957-963.	6.3	250
26	GATA6 haploinsufficiency causes pancreatic agenesis in humans. Nature Genetics, 2012, 44, 20-22.	9.4	249
27	Improved genetic testing for monogenic diabetes using targeted next-generation sequencing. Diabetologia, 2013, 56, 1958-1963.	2.9	248
28	Effective Treatment With Oral Sulfonylureas in Patients With Diabetes Due to Sulfonylurea Receptor 1 (SUR1) Mutations. Diabetes Care, 2008, 31, 204-209.	4.3	239
29	The development and validation of a clinical prediction model to determine the probability of MODY in patients with young-onset diabetes. Diabetologia, 2012, 55, 1265-1272.	2.9	238
30	HNF1B-associated renal and extra-renal diseaseâ€"an expanding clinical spectrum. Nature Reviews Nephrology, 2015, 11, 102-112.	4.1	237
31	HNF1B Mutations Associate with Hypomagnesemia and Renal Magnesium Wasting. Journal of the American Society of Nephrology: JASN, 2009, 20, 1123-1131.	3.0	234
32	Exome Sequencing Identifies a DYNC1H1 Mutation in a Large Pedigree with Dominant Axonal Charcot-Marie-Tooth Disease. American Journal of Human Genetics, 2011, 89, 308-312.	2.6	233
33	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
34	Mutations in KCNJ11, which encodes Kir6.2, are a common cause of diabetes diagnosed in the first 6Âmonths of life, with the phenotype determined by genotype. Diabetologia, 2006, 49, 1190-1197.	2.9	221
35	Recognition and Management of Individuals With Hyperglycemia Because of a Heterozygous Glucokinase Mutation. Diabetes Care, 2015, 38, 1383-1392.	4.3	217
36	Missense mutations in the insulin promoter factor-1 gene predispose to type 2 diabetes. Journal of Clinical Investigation, 1999, 104, R33-R39.	3.9	216

#	Article	IF	Citations
37	Prospective functional classification of all possible missense variants in PPARG. Nature Genetics, 2016, 48, 1570-1575.	9.4	210
38	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, 170-180.	1.1	209
39	A genetic diagnosis of HNF1A diabetes alters treatment and improves glycaemic control in the majority of insulinâ€treated patients. Diabetic Medicine, 2009, 26, 437-441.	1.2	205
40	Molecular genetics and phenotypic characteristics of MODY caused by hepatocyte nuclear factor 4Î± mutations in a large European collection. Diabetologia, 2005, 48, 878-885.	2.9	203
41	A heterozygous activating mutation in the sulphonylurea receptor SUR1 (ABCC8) causes neonatal diabetes. Human Molecular Genetics, 2006, 15, 1793-1800.	1.4	196
42	Permanent Neonatal Diabetes Caused by Dominant, Recessive, or Compound Heterozygous SUR1 Mutations with Opposite Functional Effects. American Journal of Human Genetics, 2007, 81, 375-382.	2.6	194
43	Clinical and molecular characterisation of 300 patients with congenital hyperinsulinism. European Journal of Endocrinology, 2013, 168, 557-564.	1.9	190
44	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, 3105-3110.	3.3	185
45	Relapsing diabetes can result from moderately activating mutations in KCNJ11. Human Molecular Genetics, 2005, 14, 925-934.	1.4	184
46	A high prevalence of glucokinase mutations in gestational diabetic subjects selected by clinical criteria. Diabetologia, 2000, 43, 250-253.	2.9	183
47	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, 669-685.	1.1	182
48	Islet autoantibodies can discriminate maturityâ€onset diabetes of the young (MODY) from Typeâ€ƒ1 diabetes. Diabetic Medicine, 2011, 28, 1028-1033.	1.2	173
49	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
50	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, 544-552.	2.6	171
51	The diagnosis and management of monogenic diabetes in children and adolescents. Pediatric Diabetes, 2014, 15, 47-64.	1.2	170
52	Cross-sectional and longitudinal studies suggest pharmacological treatment used in patients with glucokinase mutations does not alter glycaemia. Diabetologia, 2014, 57, 54-56.	2.9	164
53	Abnormal nephron development associated with a frameshift mutation in the transcription factor hepatocyte nuclear factor-1Î²1. Kidney International, 2000, 57, 898-907.	2.6	162
54	Mutated MESP2 Causes Spondylocostal Dysostosis in Humans. American Journal of Human Genetics, 2004, 74, 1249-1254.	2.6	157

#	Article	IF	Citations
55	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, 854-869.	1.1	157
56	Characterization of aryl hydrocarbon receptor interacting protein (AIP) mutations in familial isolated pituitary adenoma families. Human Mutation, 2010, 31, 950-960.	1.1	154
57	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, 1206-1212.	4.3	153
58	Human CHN1 Mutations Hyperactivate Î±2-Chimaerin and Cause Duane's Retraction Syndrome. Science, 2008, 321, 839-843.	6.0	152
59	AIPMutation in Pituitary Adenomas in the 18th Century and Today. New England Journal of Medicine, 2011, 364, 43-50.	13.9	151
60	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. Nature Genetics, 2013, 45, 947-950.	9.4	151
61	Insights Into the Biochemical and Genetic Basis of Glucokinase Activation From Naturally Occurring Hypoglycemia Mutations. Diabetes, 2003, 52, 2433-2440.	0.3	150
62	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
63	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-E541.	1.8	145
64	Landscape of Familial Isolated and Young-Onset Pituitary Adenomas: Prospective Diagnosis inAIPMutation Carriers. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1242-E1254.	1.8	144
65	Homozygous Mutations in NEUROD1 Are Responsible for a Novel Syndrome of Permanent Neonatal Diabetes and Neurological Abnormalities. Diabetes, 2010, 59, 2326-2331.	0.3	143
66	Atypical familial juvenile hyperuricemic nephropathy associated with a hepatocyte nuclear factor-1Î² gene mutation. Kidney International, 2003, 63, 1645-1651.	2.6	142
67	Hepatocyte nuclear factor-1 beta mutations cause neonatal diabetes and intrauterine growth retardation: support for a critical role of HNF-1? in human pancreatic development. Diabetic Medicine, 2006, 23, 1301-1306.	1.2	142
68	Heterozygous ABCC8 mutations are a cause of MODY. Diabetologia, 2012, 55, 123-127.	2.9	141
69	Permanent Neonatal Diabetes and Enteric Anendocrinosis Associated With Biallelic Mutations in NEUROG3. Diabetes, 2011, 60, 1349-1353.	0.3	138
70	KCNJ11 activating mutations are associated with developmental delay, epilepsy and neonatal diabetes syndrome and other neurological features. European Journal of Human Genetics, 2006, 14, 824-830.	1.4	134
71	Solitary functioning kidney and diverse genital tract malformations associated with hepatocyte nuclear factor-1Î² mutations. Kidney International, 2002, 61, 1243-1251.	2.6	133
72	Persistent Hyperinsulinemic Hypoglycemia and Maturity-Onset Diabetes of the Young Due to Heterozygous HNF4A Mutations. Diabetes, 2008, 57, 1659-1663.	0.3	133

#	Article	IF	Citations
73	Maturity onset diabetes of the young: identification and diagnosis. Annals of Clinical Biochemistry, 2013, 50, 403-415.	0.8	131
74	GATA6 Mutations Cause a Broad Phenotypic Spectrum of Diabetes From Pancreatic Agenesis to Adult-Onset Diabetes Without Exocrine Insufficiency. Diabetes, 2013, 62, 993-997.	0.3	128
75	Wolcott-Rallison Syndrome Is the Most Common Genetic Cause of Permanent Neonatal Diabetes in Consanguineous Families. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 4162-4170.	1.8	127
76	Urinary C-Peptide Creatinine Ratio Is a Practical Outpatient Tool for Identifying Hepatocyte Nuclear Factor 1-Î±/Hepatocyte Nuclear Factor 4-Î± Maturity-Onset Diabetes of the Young From Long-Duration Type 1 Diabetes. Diabetes Care, 2011, 34, 286-291.	4.3	123
77	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, 146-154.	7.2	123
78	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, 1230-1236.	4.3	122
79	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, 1683-1685.	2.9	120
80	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
81	Sirolimus Therapy in Infants with Severe Hyperinsulinemic Hypoglycemia. New England Journal of Medicine, 2014, 370, 1131-1137.	13.9	116
82	No Deterioration in Glycemic Control in HNF-1Â Maturity-Onset Diabetes of the Young Following Transfer From Long-Term Insulin to Sulphonylureas. Diabetes Care, 2003, 26, 3191-3192.	4.3	115
83	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, 2216-2224.	1.4	115
84	Contrasting Diabetes Phenotypes Associated With Hepatocyte Nuclear Factor-1Â and -1Â Mutations. Diabetes Care, 2004, 27, 1102-1107.	4.3	114
85	SLC2A2 mutations can cause neonatal diabetes, suggesting GLUT2 may have a role in human insulin secretion. Diabetologia, 2012, 55, 2381-2385.	2.9	113
86	Clinical presentation of 6q24 transient neonatal diabetes mellitus (6q24 TNDM) and genotypeâ€“phenotype correlation in an international cohort of patients. Diabetologia, 2013, 56, 758-762.	2.9	113
87	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
88	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
89	Hepatocyte Nuclear Factor-1Î². Journal of the American Society of Nephrology: JASN, 2001, 12, 2175-2180.	3.0	110
90	HLA Genotyping Supports a Nonautoimmune Etiology in Patients Diagnosed With Diabetes Under the Age of 6 Months. Diabetes, 2006, 55, 1895-1898.	0.3	109

#	Article	IF	Citations
91	Hepatocyte nuclear factor 1 alpha (HNF-1?) mutations in maturity-onset diabetes of the young. Human Mutation, 2000, 16, 377-385.	1.1	108
92	Â-Cell Dysfunction, Insulin Sensitivity, and Glycosuria Precede Diabetes in Hepatocyte Nuclear Factor-1Â Mutation Carriers. Diabetes Care, 2005, 28, 1751-1756.	4.3	108
93	Pregnancy outcome in patients with raised blood glucose due to a heterozygous glucokinase gene mutation. Diabetic Medicine, 2009, 26, 14-18.	1.2	108
94	GATA4 Mutations Are a Cause of Neonatal and Childhood-Onset Diabetes. Diabetes, 2014, 63, 2888-2894.	0.3	108
95	Mutations in the genes encoding the pancreatic beta-cell KATPchannel subunits Kir6.2 (KCNJ11) and SUR1 (ABCC8) in diabetes mellitus and hyperinsulinism. Human Mutation, 2006, 27, 220-231.	1.1	105
96	Clinical Heterogeneity in Patients With FOXP3 Mutations Presenting With Permanent Neonatal Diabetes. Diabetes Care, 2009, 32, 111-116.	4.3	104
97	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
98	tRNA Methyltransferase Homolog Gene TRMT10A Mutation in Young Onset Diabetes and Primary Microcephaly in Humans. PLoS Genetics, 2013, 9, e1003888.	1.5	103
99	Intrauterine Hyperglycemia Is Associated With an Earlier Diagnosis of Diabetes in HNF-1Â Gene Mutation Carriers. Diabetes Care, 2002, 25, 2287-2291.	4.3	102
100	Use of HbA1c in the Identification of Patients with Hyperglycaemia Caused by a Glucokinase Mutation: Observational Case Control Studies. PLoS ONE, 2013, 8, e65326.	1.1	101
101	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, 1276-1282.	1.8	100
102	Extreme phenotypic diversity and nonpenetrance in families with the LMNA gene mutation R644C. American Journal of Medical Genetics, Part A, 2008, 146A, 1530-1542.	0.7	100
103	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
104	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99
105	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.	1.9	97
106	Increased allâ€cause and cardiovascular mortality in monogenic diabetes as a result of mutations in the HNF1A gene. Diabetic Medicine, 2010, 27, 157-161.	1.2	96
107	Hepatocyte nuclear factor-1Â gene deletions--a common cause of renal disease. Nephrology Dialysis Transplantation, 2007, 23, 627-635.	0.4	95
108	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. Nature Communications, 2017, 8, 888.	5.8	95

#	Article	IF	Citations
109	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	2.8	93
110	Detection of an MEN1 gene mutation depends on clinical features and supports current referral criteria for diagnostic molecular genetic testing. Clinical Endocrinology, 2005, 62, 169-175.	1.2	91
111	High-Sensitivity CRP Discriminates HNF1A-MODY From Other Subtypes of Diabetes. Diabetes Care, 2011, 34, 1860-1862.	4.3	90
112	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
113	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
114	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	2.6	88
115	Permanent Neonatal Diabetes due to Paternal Germline Mosaicism for an Activating Mutation of the KCNJ11 Gene Encoding the Kir6.2 Subunit of the Î²-Cell Potassium Adenosine Triphosphate Channel. Journal of Clinical Endocrinology and Metabolism, 2004, 89, 3932-3935.	1.8	87
116	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, 14105-14113.	1.6	87
117	Diabetes Mellitus in Neonates and Infants: Genetic Heterogeneity, Clinical Approach to Diagnosis, and Therapeutic Options. Hormone Research in Paediatrics, 2013, 80, 137-146.	0.8	87
118	Novel GLIS3 mutations demonstrate an extended multisystem phenotype. European Journal of Endocrinology, 2011, 164, 437-443.	1.9	86
119	Identifying Hepatic Nuclear Factor 1Â Mutations in Children and Young Adults With a Clinical Diagnosis of Type 1 Diabetes. Diabetes Care, 2003, 26, 333-337.	4.3	84
120	Entities and frequency of neonatal diabetes: data from the diabetes documentation and quality management system (DPV). Diabetic Medicine, 2010, 27, 709-712.	1.2	84
121	Incidence, genetics, and clinical phenotype of permanent neonatal diabetes mellitus in northwest Saudi Arabia. Pediatric Diabetes, 2012, 13, 499-505.	1.2	84
122	Concordance of assays designed for the quantification of JAK2V617F: a multicenter study. Haematologica, 2009, 94, 38-45.	1.7	82
123	TheHNF4AR76W mutation causes atypical dominant Fanconi syndrome in addition to a Î² cell phenotype. Journal of Medical Genetics, 2014, 51, 165-169.	1.5	82
124	A Gene for Autosomal Recessive Spondylocostal Dysostosis Maps to 19q13.1-q13.3. American Journal of Human Genetics, 1999, 65, 175-182.	2.6	81
125	Hyperinsulinismâ€“hyperammonaemia syndrome: novel mutations in the GLUD1 gene and genotypeâ€“phenotype correlations. European Journal of Endocrinology, 2009, 161, 731-735.	1.9	81
126	Mutations in the MESP2 Gene Cause Spondylothoracic Dysostosis/Jarcho-Levin Syndrome. American Journal of Human Genetics, 2008, 82, 1334-1341.	2.6	79

#	Article	IF	Citations
127	Distinct Molecular and Morphogenetic Properties of Mutations in the Human HNF1Î² Gene That Lead to Defective Kidney Development. Journal of the American Society of Nephrology: JASN, 2003, 14, 2033-2041.	3.0	78
128	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, 758-759.	4.3	77
129	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
130	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, 131-136.	2.6	76
131	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, 1029-1031.	2.9	75
132	Micronucleus assays using cytochalasin-blocked MCL-5 cells, a proprietary human cell line expressing five human cytochromesP-450 and microsomal epoxide hydrolase. Mutagenesis, 1993, 8, 363-372.	1.0	74
133	The use of genetically engineered V79 Chinese hamster cultures expressing rat liver CYP1A1, 1A2 and 2B1 cDNAs in micronucleus assays. Mutagenesis, 1991, 6, 461-470.	1.0	73
134	Permanent neonatal diabetes due to activating mutations in ABCC8 and KCNJ11. Reviews in Endocrine and Metabolic Disorders, 2010, 11, 193-198.	2.6	73
135	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, 4695-4700.	3.3	72
136	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, 2221-2225.	1.8	72
137	Growth in PHEX-associated X-linked hypophosphatemic rickets: the importance of early treatment. Pediatric Nephrology, 2012, 27, 581-588.	0.9	71
138	C282Y mutation in HFE (haemochromatosis) gene and type 2 diabetes. Lancet, The, 1998, 351, 1933-1934.	6.3	68
139	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
140	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
141	Expanding the Clinical Spectrum Associated WithGLIS3Mutations. Journal of Clinical Endocrinology and Metabolism, 2015, 100, E1362-E1369.	1.8	66
142	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
143	Mutations at the Same Residue (R50) of Kir6.2 (KCNJ11) That Cause Neonatal Diabetes Produce Different Functional Effects. Diabetes, 2006, 55, 1705-1712.	0.3	64
144	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, 1745-1752.	0.3	64

#	Article	IF	Citations
145	Chromosome 17q12 microdeletions but not intragenic HNF1B mutations link developmental kidney disease and psychiatric disorder. Kidney International, 2016, 90, 203-211.	2.6	64
146	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	1.1	64
147	Pancreatic Endocrine and Exocrine Function in Children following Near-Total Pancreatectomy for Diffuse Congenital Hyperinsulinism. PLoS ONE, 2014, 9, e98054.	1.1	63
148	Response to treatment with rosiglitazone in familial partial lipodystrophy due to a mutation in the LMNA gene. Diabetic Medicine, 2003, 20, 823-827.	1.2	62
149	A Genome-Wide Scan in Families With Maturity-Onset Diabetes of the Young: Evidence for Further Genetic Heterogeneity. Diabetes, 2003, 52, 872-881.	0.3	62
150	Pitfalls of haplotype phasing from amplicon-based long-read sequencing. Scientific Reports, 2016, 6, 21746.	1.6	62
151	Mutations in the VNTR of the carboxyl-ester lipase gene (CEL) are a rare cause of monogenic diabetes. Human Genetics, 2010, 127, 55-64.	1.8	61
152	Childhood presentation of COL4A1 mutations. Developmental Medicine and Child Neurology, 2012, 54, 569-574.	1.1	61
153	Predictive genetic testing in maturity-onset diabetes of the young (MODY). Diabetic Medicine, 2001, 18, 417-421.	1.2	60
154	Partial and whole gene deletion mutations of the GCK and HNF1A genes in maturity-onset diabetes of the young. Diabetologia, 2007, 50, 2313-2317.	2.9	59
155	Recessively InheritedLRBAMutations Cause Autoimmunity Presenting as Neonatal Diabetes. Diabetes, 2017, 66, 2316-2322.	0.3	59
156	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, 636-638.	1.8	58
157	An ABCC8 Gene Mutation and Mosaic Uniparental Isodisomy Resulting in Atypical Diffuse Congenital Hyperinsulinism. Diabetes, 2008, 57, 259-263.	0.3	58
158	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, 2020, 130, 6338-6353.	3.9	58
159	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, 859-862.	2.9	57
160	KATP channel mutations in infants with permanent diabetes diagnosed after 6 months of life. Pediatric Diabetes, 2012, 13, 322-325.	1.2	57
161	Recessive SLC19A2 mutations are a cause ofÂneonatal diabetes mellitus inÂthiamine-responsive megaloblastic anaemia. Pediatric Diabetes, 2012, 13, 314-321.	1.2	57
162	The Generalized Aminoaciduria Seen in Patients With Hepatocyte Nuclear Factor-1Â Mutations Is a Feature of All Patients With Diabetes and Is Associated With Glucosuria. Diabetes, 2001, 50, 2047-2052.	0.3	56

#	Article	IF	Citations
163	Etiological Investigation of Diabetes in Young Adults Presenting With Apparent Type 2 Diabetes. Diabetes Care, 2003, 26, 2088-2093.	4.3	56
164	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's anaemia cells. Carcinogenesis, 1993, 14, 2487-2494.	1.3	54
165	Permanent neonatal diabetes mellitus: prevalence and genetic diagnosis in the SEARCH for Diabetes in Youth Study. Pediatric Diabetes, 2012, 14, n/a-n/a.	1.2	54
166	An investigation into the activation and deactivation of chlorinated hydrocarbons to genotoxins in metabolically competent human cells. Mutagenesis, 1996, 11, 247-274.	1.0	53
167	Long-Term Follow-Up of Children With Congenital Hyperinsulinism on Octreotide Therapy. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 3660-3667.	1.8	53
168	Origin of de Novo KCNJ11 Mutations and Risk of Neonatal Diabetes for Subsequent Siblings. Journal of Clinical Endocrinology and Metabolism, 2007, 92, 1773-1777.	1.8	52
169	Incidence of neonatal diabetes in Austria-calculation based on the Austrian Diabetes Register. Pediatric Diabetes, 2010, 11, 18-23.	1.2	52
170	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.	1.9	52
171	Frequent Occurrence of an Intron 4 Mutation in Multiple Endocrine Neoplasia Type 1. Journal of Clinical Endocrinology and Metabolism, 2002, 87, 2688-2693.	1.8	51
172	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.	0.3	51
173	Permanent neonatal diabetes in an Asian infant. Journal of Pediatrics, 2005, 146, 131-133.	0.9	51
174	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, 18988-18992.	3.3	51
175	Persistently autoantibody negative (PAN) type 1 diabetes mellitus in children. Pediatric Diabetes, 2011, 12, 142-149.	1.2	51
176	Genome-Wide Homozygosity Analysis Reveals HADH Mutations as a Common Cause of Diazoxide-Responsive Hyperinsulinemic-Hypoglycemia in Consanguineous Pedigrees. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E498-E502.	1.8	51
177	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	1.4	51
178	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. Brain, 2019, 142, 50-58.	3.7	51
179	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, 1152-1154.	0.3	50
180	Messenger RNA Transcripts of the Hepatocyte Nuclear Factor-1Â Gene Containing Premature Termination Codons Are Subject to Nonsense-Mediated Decay. Diabetes, 2004, 53, 500-504.	0.3	50

#	Article	IF	Citations
181	Antenatal Diagnosis of Fetal Genotype Determines if Maternal Hyperglycemia Due to a Glucokinase Mutation Requires Treatment. Diabetes Care, 2012, 35, 1832-1834.	4.3	50
182	Mutations in the hepatocyte nuclear factor-1Î² (HNF1B) gene are common with combined uterine and renal malformations but are not found with isolated uterine malformations. American Journal of Obstetrics and Gynecology, 2010, 203, 364.e1-364.e5.	0.7	49
183	Three novel mutations in KIF21A highlight the importance of the third coiled-coil stalk domain in the etiology of CFEOM1. BMC Genetics, 2007, 8, 26.	2.7	48
184	SomaticGPR101Duplication Causing X-Linked Acrogigantism (XLAG)â€"Diagnosis and Management. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 1927-1930.	1.8	48
185	Premature birth and low birth weight associated with nonautoimmune hyperthyroidism due to an activating thyrotropin receptor gene mutation. Clinical Endocrinology, 2004, 60, 711-718.	1.2	47
186	Permanent Neonatal Diabetes Mellitus Caused by a Novel Homozygous (T168A) Glucokinase (GCK) Mutation: Initial Response to Oral Sulphonylurea Therapy. Journal of Pediatrics, 2008, 153, 122-126.	0.9	46
187	Biallelic PDX1 (insulin promoter factorÂ1) mutations causing neonatal diabetes without exocrine pancreatic insufficiency. Diabetic Medicine, 2013, 30, e197-200.	1.2	46
188	The Common p.R114W HNF4A Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. Diabetes, 2016, 65, 3212-3217.	0.3	46
189	Prevalence of diabetes in Australia: insights from the Fremantle Diabetes Study Phase II. Internal Medicine Journal, 2018, 48, 803-809.	0.5	46
190	Parent-offspring trios: a resource to facilitate the identification of type 2 diabetes genes. Diabetes, 1999, 48, 2475-2479.	0.3	45
191	Heterogeneity in young adult onset diabetes: aetiology alters clinical characteristics. Diabetic Medicine, 2002, 19, 758-761.	1.2	45
192	The position of premature termination codons in the hepatocyte nuclear factor â^'1 beta gene determines susceptibility to nonsense-mediated decay. Human Genetics, 2005, 118, 214-224.	1.8	45
193	Clinical and molecular characterisation of hyperinsulinaemic hypoglycaemia in infants born small-for-gestational age. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2013, 98, F356-F358.	1.4	44
194	Characteristics of maturity onset diabetes of the young in a large diabetes center. Pediatric Diabetes, 2016, 17, 360-367.	1.2	44
195	Next generation sequencing of chromosomal rearrangements in patients with split-hand/split-foot malformation provides evidence forDYNC1I1exonic enhancers ofDLX5/6expression in humans. Journal of Medical Genetics, 2014, 51, 264-267.	1.5	43
196	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
197	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
198	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

#	Article	IF	Citations
199	40th EASD Annual Meeting of the European Association for the Study of Diabetes. Diabetologia, 2004, 47, A1-A464.	2.9	41
200	Leucine-sensitive hyperinsulinaemic hypoglycaemia in patients with loss of function mutations in 3-Hydroxyacyl-CoA Dehydrogenase. Orphanet Journal of Rare Diseases, 2012, 7, 25.	1.2	41
201	Phenotypic severity of homozygous GCK mutations causing neonatal or childhood-onset diabetes is primarily mediated through effects on protein stability. Human Molecular Genetics, 2014, 23, 6432-6440.	1.4	41
202	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
203	ACE Gene Polymorphism as a Prognostic Indicator in Patients With Type 2 Diabetes and Established Renal Disease. Diabetes Care, 2001, 24, 2115-2120.	4.3	40
204	Permanent neonatal diabetes: different aetiology in Arabs compared to Europeans. Archives of Disease in Childhood, 2012, 97, 721-723.	1.0	40
205	Whole gene deletion of the hepatocyte nuclear factor-1Â gene in a patient with the prune-belly syndrome. Nephrology Dialysis Transplantation, 2008, 23, 2412-2415.	0.4	39
206	Clinical characterization of a newly described neonatal diabetes syndrome caused by RFX6 mutations. American Journal of Medical Genetics, Part A, 2011, 155, 2821-2825.	0.7	39
207	Lipoprotein composition in HNF1A-MODY: Differentiating between HNF1A-MODY and Type 2 diabetes. Clinica Chimica Acta, 2012, 413, 927-932.	0.5	39
208	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
209	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
210	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
211	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.4	39
212	A further example of a distinctive autosomal recessive syndrome comprising neonatal diabetes mellitus, intestinal atresias and gall bladder agenesis. American Journal of Medical Genetics, Part A, 2008, 146A, 1713-1717.	0.7	38
213	Abnormal splicing of hepatocyte nuclear factor-1 beta in the renal cysts and diabetes syndrome. Diabetologia, 2004, 47, 937-942.	2.9	37
214	Analysis of gross deletions in the MEN1 gene in patients with multiple endocrine neoplasia typeÂ1. Clinical Endocrinology, 2008, 68, 350-354.	1.2	37
215	Polygenic Risk Variants for Type 2 Diabetes Susceptibility Modify Age at Diagnosis in Monogenic HNF1A Diabetes. Diabetes, 2010, 59, 266-271.	0.3	37
216	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

#	Article	IF	Citations
217	Adjacent mutations in the gating loop of Kir6.2 produce neonatal diabetes and hyperinsulinism. EMBO Molecular Medicine, 2009, 1, 166-177.	3.3	36
218	Clinical and histological heterogeneity of congenital hyperinsulinism due to paternally inherited heterozygous ABCC8/KCNJ11 mutations. European Journal of Endocrinology, 2014, 171, 685-695.	1.9	36
219	Mutations in the ABCC8 (SUR1 subunit of the K_{ATP} channel) gene are associated with a variable clinical phenotype. Clinical Endocrinology, 2009, 71, 358-362.	1.2	35
220	Risk category system to identify pituitary adenoma patients with AIP mutations. Journal of Medical Genetics, 2018, 55, 254-260.	1.5	35
221	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
222	Identification of a Novel Î²-Cell Glucokinase (GCK) Promoter Mutation (âˆ'71G>C) That ModulatesGCKGene Expression Through Loss of Allele-Specific Sp1 Binding Causing Mild Fasting Hyperglycemia in Humans. Diabetes, 2009, 58, 1929-1935.	0.3	34
223	Mutation of HES7 in a large extended family with spondylocostal dysostosis and dextrocardia with situs inversus. American Journal of Medical Genetics, Part A, 2013, 161, 2244-2249.	0.7	34
224	HNF1B deletions in patients with youngâ€onset diabetes but no known renal disease. Diabetic Medicine, 2013, 30, 114-117.	1.2	34
225	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. European Journal of Human Genetics, 2015, 23, 1744-1748.	1.4	34
226	Neonatal diabetes in Ukraine: incidence, genetics, clinical phenotype and treatment. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 1279-86.	0.4	34
227	A modified protocol for the cytochalasin B in vitro micronucleus assay using whole human blood or separated lymphocyte cultures. Mutagenesis, 1993, 8, 317-320.	1.0	33
228	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
229	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
230	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
231	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. Diabetes, 1999, 48, 921-923.	0.3	32
232	Activating AKT2 Mutation: Hypoinsulinemic Hypoketotic Hypoglycemia. Journal of Clinical Endocrinology and Metabolism, 2014, 99, 391-394.	1.8	32
233	Noninvasive Fetal Genotyping by Droplet Digital PCR to Identify Maternally Inherited Monogenic Diabetes Variants. Clinical Chemistry, 2020, 66, 958-965.	1.5	32
234	Severe Intrauterine Growth Retardation and Atypical Diabetes Associated with a Translocation Breakpoint Disrupting Regulation of the Insulin-Like Growth Factor 2 Gene. Journal of Clinical Endocrinology and Metabolism, 2008, 93, 4373-4380.	1.8	31

#	Article	IF	Citations
235	Familial Focal Congenital Hyperinsulinism. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 24-28.	1.8	31
236	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
237	Genomic variant sharing: a position statement. Wellcome Open Research, 2019, 4, 22.	0.9	31
238	Use of multicolour chromosome painting to identify chromosomal rearrangements in human lymphocytes exposed to bleomycin: A comparison with conventional cytogenetic analysis of giemsa-stained chromosomes. Environmental and Molecular Mutagenesis, 1995, 26, 44-54.	0.9	30
239	Glucokinase mutations in a phenotypically selected multiethnic group of women with a history of gestational diabetes. Diabetic Medicine, 2001, 18, 683-684.	1.2	30
240	A family with a novel TSH receptor activating germline mutation (p.Ala485Val). European Journal of Pediatrics, 2008, 167, 1231-1237.	1.3	30
241	Mutations in the Glucokinase Gene of the Fetus Result in Reduced Placental Weight. Diabetes Care, 2008, 31, 753-757.	4.3	30
242	Liver Disease and Other Comorbidities in Wolcott-Rallison Syndrome: Different Phenotype and Variable Associations in a Large Cohort. Hormone Research in Paediatrics, 2015, 83, 190-197.	0.8	30
243	Cancer Variant Interpretation Group UK (CanVIG-UK): an exemplar national subspecialty multidisciplinary network. Journal of Medical Genetics, 2020, 57, 829-834.	1.5	30
244	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. Human Genetics, 1997, 101, 351-354.	1.8	29
245	Sulfonylurea Treatment in Young Children With Neonatal Diabetes: Dealing with hyperglycemia, hypoglycemia, and sick days. Diabetes Care, 2007, 30, e28-e29.	4.3	29
246	Patients' understanding of genetic susceptibility testing in mainstream medicine: qualitative study on thrombophilia. BMC Health Services Research, 2007, 7, 82.	0.9	29
247	A Kir6.2 mutation causing severe functional effects in vitro produces neonatal diabetes without the expected neurological complications. Diabetologia, 2008, 51, 802-810.	2.9	29
248	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
249	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
250	Evidence for Haploinsufficiency of the Human HNF1Î± Gene Revealed by Functional Characterization of MODY3-Associated Mutations. Biological Chemistry, 2002, 383, 1691-700.	1.2	28
251	Rapid and Sensitive Real-Time Polymerase Chain Reaction Method for Detection and Quantification of 3243A>G Mitochondrial Point Mutation. Journal of Molecular Diagnostics, 2006, 8, 225-230.	1.2	28
252	Characterization of ABCC8 and KCNJ11 gene mutations and phenotypes in Korean patients with congenital hyperinsulinism. European Journal of Endocrinology, 2011, 164, 919-926.	1.9	28

#	Article	IF	Citations
253	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
254	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
255	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
256	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-4Â (HNF4A) Gene. Diabetes, 2002, 51, 2329-2333.	0.3	27
257	A Comparison of Methods for EGFR Mutation Testing in Nonâ€“Small Cell Lung Cancer. Diagnostic Molecular Pathology, 2013, 22, 190-195.	2.1	27
258	Neurogenin 3 is important but not essential for pancreatic islet development in humans. Diabetologia, 2014, 57, 2421-2424.	2.9	27
259	Prematurity and Genetic Testing for Neonatal Diabetes. Pediatrics, 2016, 138, .	1.0	27
260	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
261	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.	3.9	27
262	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
263	Genetic testing for glucokinase mutations in clinically selected patients with MODY: a worthwhile investment. Swiss Medical Weekly, 2005, 135, 352-6.	0.8	27
264	Mutational analyses of UPIIIA, SHH, EFNB2, and HNF1Î² in persistent cloaca and associated kidney malformations. Journal of Pediatric Urology, 2007, 3, 2-9.	0.6	26
265	Transcription factor HNF1Î² and novel partners affect nephrogenesis. Kidney International, 2008, 74, 210-217.	2.6	26
266	Diabetes Susceptibility in the Canadian Oji-Cree Population Is Moderated by Abnormal mRNA Processing of HNF1A G319S Transcripts. Diabetes, 2008, 57, 1978-1982.	0.3	26
267	Cushing Syndrome Secondary to A Thymic Carcinoid Tumor Due to Multiple Endocrine Neoplasia Type 1. Endocrine Practice, 2011, 17, e92-e96.	1.1	26
268	The Heterogeneity of Focal Forms of Congenital Hyperinsulinism. Journal of Clinical Endocrinology and Metabolism, 2012, 97, E94-E99.	1.8	26
269	Clinical utility gene card for: Maturity-onset diabetes of the young. European Journal of Human Genetics, 2014, 22, 1153-1153.	1.4	26
270	Pharmacogenomics in diabetes: outcomes of thiamine therapy in TRMA syndrome. Diabetologia, 2018, 61, 1027-1036.	2.9	26

#	Article	IF	Citations
271	The expression of cytochrome P450IIB1 in Saccharomyces cerevisia results in an increased mutation frequency when exposed to cyclophosphamide. Carcinogenesis, 1989, 10, 2139-2143.	1.3	25
272	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. Diabetes, 2011, 60, 1813-1822.	0.3	25
273	Thiamine responsive megaloblastic anemia: a novelSLC19A2compound heterozygous mutation in two siblings. Pediatric Diabetes, 2013, 14, 384-387.	1.2	25
274	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
275	Increased Population Risk ofAIP-Related Acromegaly and Gigantism in Ireland. Human Mutation, 2017, 38, 78-85.	1.1	25
276	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
277	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
278	Development of a Quantitative Real-Time Polymerase Chain Reaction Assay for the Detection of the JAK2 V617F Mutation. Journal of Molecular Diagnostics, 2007, 9, 42-46.	1.2	24
279	Hypoglycaemia following diabetes remission in patients with 6q24 methylation defects: expanding the clinical phenotype. Diabetologia, 2013, 56, 218-221.	2.9	24
280	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
281	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
282	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
283	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
284	Severe hyperglycemia after renal transplantation in a pediatric patient with a mutation of the Hepatocyte Nuclear Factor-1Î² gene. American Journal of Kidney Diseases, 2002, 40, 1325-1330.	2.1	23
285	Abnormal splicing of hepatocyte nuclear factor 1 alpha in maturity-onset diabetes of the young. Diabetologia, 2002, 45, 1463-1467.	2.9	23
286	Apolipoprotein-E Influences Aspects of Intellectual Ability in Type 1 Diabetes. Diabetes, 2003, 52, 145-148.	0.3	23
287	Severe Insulin Resistance and Intrauterine Growth Deficiency Associated With Haploinsufficiency forINSRandCHN2. Diabetes, 2009, 58, 2954-2961.	0.3	23
288	Congenital Hyperinsulinism due to a Compound Heterozygous <i>ABCC8 </i>Mutation with Spontaneous Resolution at Eight Weeks. Hormone Research in Paediatrics, 2010, 73, 287-292.	0.8	23

#	Article	IF	Citations
289	Identification of a SLC19A2 nonsense mutation in Persian families with thiamine-responsive megaloblastic anemia. Gene, 2013, 519, 295-297.	1.0	23
290	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
291	Asian MODY: are we missing an important diagnosis?. Diabetic Medicine, 2006, 23, 1257-1260.	1.2	22
292	Defining the genetic aetiology of monogenic diabetes can improve treatment. Expert Opinion on Pharmacotherapy, 2006, 7, 1759-1767.	0.9	22
293	In vitro expression of NGN3 identifies RAB3B as the predominant Ras-associated GTP-binding protein 3 family member in human islets. Journal of Endocrinology, 2010, 207, 151-161.	1.2	22
294	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. Diabetes, 2016, 65, 2810-2815.	0.3	22
295	Permanent neonatal diabetes mellitus and neurological abnormalities due to a novel homozygous missense mutation inNEUROD1. Pediatric Diabetes, 2018, 19, 898-904.	1.2	22
296	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. Diabetes, 2019, 68, 1528-1535.	0.3	22
297	Homozygosity mapping provides supporting evidence of pathogenicity in recessive Mendelian disease. Genetics in Medicine, 2019, 21, 982-986.	1.1	22
298	Mesangiocapillary Glomerulonephritis Type 2 Associated with Familial Partial Lipodystrophy (Dunnigan-Kobberling Syndrome). Nephron Clinical Practice, 2004, 96, c35-c38.	2.3	21
299	Discovery of a Novel Site Regulating Glucokinase Activity following Characterization of a New Mutation Causing Hyperinsulinemic Hypoglycemia in Humans. Journal of Biological Chemistry, 2011, 286, 19118-19126.	1.6	21
300	<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
301	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
302	A comparative study of the use of primary Chinese hamster liver cultures and genetically engineered immortal V79 Chinese hamster cell lines expressing rat liver CYP1A1, 1A2 and 2B1 cDNAs in micronucleus assays. Toxicology, 1993, 82, 131-149.	2.0	20
303	Glibenclamide unresponsiveness in a Brazilian child with permanent neonatal diabetes mellitus and DEND syndrome due to a C166Y mutation in KCNJ11 (Kir6.2) gene. Arquivos Brasileiros De Endocrinologia E Metabologia, 2008, 52, 1350-1355.	1.3	20
304	Germline mutations in the CDKN1B gene encoding p27^{Kip1} are a rare cause of multiple endocrine neoplasia type 1. Clinical Endocrinology, 2009, 70, 499-500.	1.2	20
305	GeneScreen: a program for high-throughput mutation detection in DNA sequence electropherograms. Journal of Medical Genetics, 2011, 48, 123-130.	1.5	20
306	A novel GATA6 mutation leading to congenital heart defects and permanent neonatal diabetes: A case report. Diabetes and Metabolism, 2013, 39, 370-374.	1.4	20

#	Article	IF	Citations
307	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.	0.4	20
308	AIP mutations in young patients with acromegaly and the Tampico Giant: the Mexican experience. Endocrine, 2016, 53, 402-411.	1.1	20
309	A conserved tryptophan at the membraneâ€“water interface acts as a gatekeeper for Kir6.2/SUR1 channels and causes neonatal diabetes when mutated. Journal of Physiology, 2011, 589, 3071-3083.	1.3	19
310	Hepatoblastoma in a child with a paternally-inherited ABCC8 mutation and mosaic paternal uniparental disomy 11p causing focal congenital hyperinsulinism. European Journal of Medical Genetics, 2013, 56, 114-117.	0.7	19
311	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.	0.4	19
312	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.	1.8	19
313	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
314	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
315	A Combination of Nifedipine and Octreotide Treatment in an Hyperinsulinemic Hypoglycemic Infant. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2014, 6, 119-121.	0.4	18
316	Differential regulation of serum microRNA expression by HNF1Î² and HNF1Î± transcription factors. Diabetologia, 2016, 59, 1463-1473.	2.9	18
317	The prevalence of monogenic diabetes in Australia: the Fremantle Diabetes Study Phase II. Medical Journal of Australia, 2017, 207, 344-347.	0.8	18
318	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	2.6	18
319	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
320	SavvyCNV: Genome-wide CNV calling from off-targetÂreads. PLoS Computational Biology, 2022, 18, e1009940.	1.5	18
321	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, 636-638.	1.8	17
322	No Evidence for Linkage at Candidate Type 2 Diabetes Susceptibility Loci on Chromosomes 12 and 20 in United Kingdom Caucasians. Journal of Clinical Endocrinology and Metabolism, 2000, 85, 853-857.	1.8	17
323	Novel Mutations in X-Linked Dominant Chondrodysplasia Punctata (CDPX2). Journal of Investigative Dermatology, 2003, 121, 939-942.	0.3	17
324	Semi-Automated Unidirectional Sequence Analysis for Mutation Detection in a Clinical Diagnostic Setting. Genetic Testing and Molecular Biomarkers, 2009, 13, 381-386.	0.3	17

#	Article	IF	Citations
325	Neonatal hyperinsulinaemic hypoglycaemia and monogenic diabetes due to a heterozygous mutation of the HNF4A gene. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2009, 49, 328-330.	0.4	17
326	Autosomal dominant spondylocostal dysostosis in three generations of a Macedonian family: Negative mutation analysis of DLL3, MESP2, HES7, and LFNG. American Journal of Medical Genetics, Part A, 2010, 152A, 1378-1382.	0.7	17
327	Clinical characteristics of recessive and dominant congenital hyperinsulinism due to mutation(s) in the ABCC8/KCNJ11 genes encoding the ATP-sensitive potasium channel in the pancreatic beta cell. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 1019-23.	0.4	17
328	In Vitro Recovery of ATP-Sensitive Potassium Channels in Â-Cells From Patients With Congenital Hyperinsulinism of Infancy. Diabetes, 2011, 60, 1223-1228.	0.3	17
329	Permanent neonatal diabetes mellitus due to KCNJ11 mutation in a Portuguese family: transition from insulin to oral sulfonylureas. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 367-70.	0.4	17
330	Identification of a novel nonsense mutation and a missense substitution in the AGPAT2 gene causing congenital generalized lipodystrophy type 1. European Journal of Medical Genetics, 2012, 55, 620-624.	0.7	17
331	Genetic studies in a coexistence of acromegaly, pheochromocytoma, gastrointestinal stromal tumor (GIST) and thyroid follicular adenoma. Arquivos Brasileiros De Endocrinologia E Metabologia, 2012, 56, 507-512.	1.3	17
332	Clinical characteristics and phenotypeâ€“genotype analysis in Turkish patients with congenital hyperinsulinism; predominance of recessive KATP channel mutations. European Journal of Endocrinology, 2014, 170, 885-892.	1.9	17
333	Fanconi-Bickel Syndrome - Mutation in SLC2A2 Gene. Indian Journal of Pediatrics, 2014, 81, 1237-1239.	0.3	17
334	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
335	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
336	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
337	An In-Frame Deletion in Kir6.2 (KCNJ11) Causing Neonatal Diabetes Reveals a Site of Interaction between Kir6.2 and SUR1. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 2551-2557.	1.8	16
338	Ten years of the national genetic diabetes nurse network: a model for the translation of genetic information into clinical care. Clinical Medicine, 2014, 14, 117-121.	0.8	16
339	Genome, Exome, and Targeted Next-Generation Sequencing in Neonatal Diabetes. Pediatric Clinics of North America, 2015, 62, 1037-1053.	0.9	16
340	Genetic mutations associated with neonatal diabetes mellitus in Omani patients. Journal of Pediatric Endocrinology and Metabolism, 2018, 31, 195-204.	0.4	16
341	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	3.6	16
342	Genetic testing for maturity onset diabetes of the young in childhood hyperglycaemia. Archives of Disease in Childhood, 1998, 78, 552-554.	1.0	15

#	Article	IF	Citations
343	Non-penetrance in a MODY 3 family with a mutation in the hepatic nuclear factor 1Î± gene: implications for predictive testing. European Journal of Human Genetics, 1999, 7, 729-732.	1.4	15
344	Autosomal dominant inheritance of non-syndromic renal hypoplasia and dysplasia: dramatic variation in clinical severity in a single kindred. Nephrology Dialysis Transplantation, 2006, 22, 259-263.	0.4	15
345	Mosaic Paternal Uniparental Isodisomy and an ABCC8 Gene Mutation in a Patient With Permanent Neonatal Diabetes and Hemihypertrophy. Diabetes, 2008, 57, 255-258.	0.3	15
346	Interaction between mutations in the slide helix of Kir6.2 associated with neonatal diabetes and neurological symptoms. Human Molecular Genetics, 2010, 19, 963-972.	1.4	15
347	The previously reported T342P GCK missense variant is not a pathogenic mutation causing MODY. Diabetologia, 2011, 54, 2202-2205.	2.9	15
348	Case Reports. Indian Pediatrics, 2011, 48, 727-736.	0.2	15
349	Transient neonatal diabetes with two novel mutations in the KCNJ11 gene and response to sulfonylurea treatment in a preterm infant. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 1077-80.	0.4	15
350	Insights Into the Pathogenicity of Rare Missense GCK Variants From the Identification and Functional Characterization of Compound Heterozygous and Double Mutations Inherited in Cis. Diabetes Care, 2012, 35, 1482-1484.	4.3	15
351	Assessment of the HNF1B Score as a Tool to Select Patients for <i>HNF1B</i> Genetic Testing. Nephron, 2015, 130, 134-140.	0.9	15
352	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.	0.4	15
353	Variable Phenotype of Diabetes Mellitus in Siblings with a Homozygous <i>PTF1A</i> Enhancer Mutation. Hormone Research in Paediatrics, 2015, 84, 206-211.	0.8	15
354	Biomonitoring study of a group of workers potentially exposed to traffic fumes. , 1997, 30, 119-130.		14
355	A severe clinical phenotype results from the co-inheritance of type 2 susceptibility genes and a hepatocyte nuclear factor-1alpha mutation. Diabetes Care, 2000, 23, 424-425.	4.3	14
356	Molecular genetic prenatal diagnosis for a case of autosomal recessive spondylocostal dysostosis. Prenatal Diagnosis, 2003, 23, 575-579.	1.1	14
357	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
358	A comparison of conventional metaphase analysis of Giemsa-stained chromosomes with multi-colour fluorescence in situ hybridization analysis to detect chromosome aberrations induced by daunomycin. Mutagenesis, 1996, 11, 537-546.	1.0	13
359	Permanent neonatal diabetes mellitus in Jordan. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 879-83.	0.4	13
360	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, 144-147.	0.4	13

#	Article	IF	Citations
361	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
362	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
363	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
364	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. Lipids in Health and Disease, 2006, 5, 11.	1.2	12
365	Digenic heterozygousHNF1AandHNF4Amutations in two siblings with childhood-onset diabetes. Pediatric Diabetes, 2013, 14, 535-538.	1.2	12
366	Screening for neonatal diabetes at day 5 of life using dried blood spot glucose measurement. Diabetologia, 2017, 60, 2168-2173.	2.9	12
367	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
368	Fainting Fanconi syndrome clarified by proxy: a case report. BMC Nephrology, 2017, 18, 230.	0.8	12
369	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
370	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
371	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
372	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
373	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. Diabetologia, 2022, 65, 336-342.	2.9	12
374	Induction of micronuclei in V79 Chinese hamster cells by hydroquinone and econazole nitrate. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 1993, 287, 87-91.	0.4	11
375	Sequencing PDX1 (insulin promoter factorâ€ƒ1) in 1788 UK individuals found 5% had a low frequency coding variant, but these variants are not associated with Typeâ€ƒ2 diabetes. Diabetic Medicine, 2011, 28, 681-684.	1.2	11
376	Variable phenotype in five patients with Wolcott-Rallison syndrome due to the same EIF2AK3 (c.1259delA) mutation. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 757-60.	0.4	11
377	<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
378	Paternal Uniparental Isodisomy of Chromosome 11p15.5 within the Pancreas Causes Isolated Hyperinsulinemic Hypoglycemia. Frontiers in Endocrinology, 2011, 2, 66.	1.5	10

#	Article	IF	Citations
379	Wolcott-Rallison Syndrome Due to a Novel Mutation (R491X) in EIF2AK3 Gene. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2012, 4, 101-103.	0.4	10
380	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
381	Genotype and phenotype correlations in Iranian patients with hyperinsulinaemic hypoglycaemia. BMC Research Notes, 2015, 8, 350.	0.6	10
382	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
383	Sirolimus-Induced Hepatitis in Two Patients with Hyperinsulinemic Hypoglycemia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 279-283.	0.4	10
384	Phenotypic Multiple Endocrine Neoplasia Type 2B, Without Endocrinopathy or RET Gene Mutation: Implications for Management. Thyroid, 2006, 16, 605-608.	2.4	9
385	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
386	A Novel KCNJ11 Mutation Associated with Transient Neonatal Diabetes. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2018, 10, 175-178.	0.4	9
387	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	1.4	9
388	Neonatal diabetes mutations disrupt a chromatin pioneering function that activates the human insulin gene. Cell Reports, 2021, 35, 108981.	2.9	9
389	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
390	Proposed mechanism for a novel insertion/deletion frameshift mutation (I414G415ATCG?CCA) in the hepatocyte nuclear factor 1 alpha (HNF-1?) gene which causes maturity-onset diabetes of the young (MODY). Human Mutation, 2000, 16, 273-273.	1.1	8
391	Observationsï¿½Maturity onset diabetes of the young (MODY) and early onset Type II diabetes are not caused by loss of imprinting at the transient neonatal diabetes (TNDM) locus. Diabetologia, 2001, 44, 924-924.	2.9	8
392	Variability in the age at diagnosis of diabetes in two unrelated patients with a homozygous glucokinase gene mutation. Journal of Pediatric Endocrinology and Metabolism, 2012, 25, 805-8.	0.4	8
393	Permanent Neonatal Diabetes Caused by a Novel Mutation. Indian Pediatrics, 2012, 49, 486-488.	0.2	8
394	Congenital hyperinsulinism: marked clinical heterogeneity in siblings with identical mutations in the ABCC8 gene. Clinical Endocrinology, 2012, 76, 312-313.	1.2	8
395	Increased Plasma Incretin Concentrations Identifies a Subset of PatientsÂwith Persistent Congenital Hyperinsulinism without KATPÂChannelÂGene Defects. Journal of Pediatrics, 2015, 166, 191-194.	0.9	8
396	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

#	Article	IF	Citations
397	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
398	Sequencing of candidate genes selected by beta cell experts in monogenic diabetes of unknown aetiology. JOP: Journal of the Pancreas, 2010, 11, 14-7.	1.5	8
399	Prenatal testing for a novelEBP missense mutation causing X-linked dominant chondrodysplasia punctata. Prenatal Diagnosis, 2008, 28, 384-388.	1.1	7
400	The association of cardiac ventricular hypertrophy with congenital hyperinsulinism. European Journal of Endocrinology, 2012, 167, 619-624.	1.9	7
401	Thiamine responsive megaloblastic anemia with a novel SLC19A2 mutation presenting with myeloid maturational arrest. Pediatric Blood and Cancer, 2013, 60, 1242-1243.	0.8	7
402	Permanent neonatal diabetes in siblings with novel C109YINSmutation transmitted by an unaffected parent with somatic mosaicism. Pediatric Diabetes, 2014, 15, 324-328.	1.2	7
403	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
404	MODY in Ukraine: genes, clinical phenotypes and treatment. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 1095-1103.	0.4	7
405	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
406	Study of Acute Liver Failure in Children Using Next Generation Sequencing Technology. Journal of Pediatrics, 2021, 236, 124-130.	0.9	7
407	Genomic variant sharing: a position statement. Wellcome Open Research, 0, 4, 22.	0.9	7
408	Misannotation of multiple-nucleotide variants risks misdiagnosis. Wellcome Open Research, 0, 4, 145.	0.9	7
409	Increased sterigmatocystin-induced mutation frequency in Saccharomyces cerevisiae expressing cytochrome P450 CYP2B1. Biochemical Pharmacology, 1992, 43, 374-376.	2.0	6
410	Prematurity, macrosomia, hyperinsulinaemic hypoglycaemia and a dominant ABCC8 gene mutation. BMJ Case Reports, 2013, 2013, bcr2013008767-bcr2013008767.	0.2	6
411	Variants in KCNJ11 and BAD do not predict response to ketogenic dietary therapies for epilepsy. Epilepsy Research, 2015, 118, 22-28.	0.8	6
412	Hyperinsulinemic hypoglycemia of infancy due to novel HADH mutation in two siblings. Indian Pediatrics, 2016, 53, 912-913.	0.2	6
413	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
414	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

#	Article	IF	Citations
415	R127W in HNF4? is a loss-of-function mutation causing maturity-onset diabetes of the young (MODY) in a UK Caucasian family. Diabetologia, 2000, 43, 1203-1203.	2.9	5
416	Mutations in HHEX are not a common cause of monogenic forms of beta cell dysfunction. Diabetologia, 2007, 50, 2019-2022.	2.9	5
417	The Renal Cysts and Diabetes (RCAD) Syndrome in a Child with Deletion of the Hepatocyte Nuclear Factor-1Î² Gene. Indian Journal of Pediatrics, 2010, 77, 1429-1431.	0.3	5
418	Focal congenital hyperinsulinism in a patient with septo-optic dysplasia. Nature Reviews Endocrinology, 2010, 6, 646-650.	4.3	5
419	KCNJ11 activating mutations cause both transient and permanent neonatal diabetes mellitus in Cypriot patients. Pediatric Diabetes, 2011, 12, 133-137.	1.2	5
420	Clinical and molecular basis of transient neonatal diabetes mellitus in Brazilian children. Diabetes Research and Clinical Practice, 2012, 97, e41-e44.	1.1	5
421	Primary hypothyroidism: an unusual manifestation of Wolcottâ€“Rallison syndrome. European Journal of Pediatrics, 2014, 173, 1565-1568.	1.3	5
422	Reclassification of Diabetes Etiology in a Family With Multiple Diabetes Phenotypes. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1067-E1071.	1.8	5
423	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
424	Focal Congenital Hyperinsulinism as a Cause for Sudden Infant Death. Pediatric and Developmental Pathology, 2019, 22, 65-69.	0.5	5
425	Refinement of the critical genomic region for congenital hyperinsulinismÂin the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	0.9	5
426	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
427	Renal cysts and diabetes due to a heterozygous HNF-1Â gene deletion. Nephrology Dialysis Transplantation, 2007, 22, 1271-1272.	0.4	4
428	Evaluation of 13q14 Status in Patients with Chronic Lymphocytic Leukemia Using Single Nucleotide Polymorphism-Based Techniques. Journal of Molecular Diagnostics, 2009, 11, 298-305.	1.2	4
429	Biochemical evaluation of an infant with hypoglycemia resulting from a novel de novo mutation of the GLUD1 gene and hyperinsulinism-hyperammonemia syndrome. Journal of Pediatric Endocrinology and Metabolism, 2011, 24, 573-7.	0.4	4
430	Amino acid properties may be useful in predicting clinical outcome in patients with Kir6.2 neonatal diabetes. European Journal of Endocrinology, 2012, 167, 417-421.	1.9	4
431	Permanent neonatal diabetes caused by a novel mutation in the INS gene. Diabetes Research and Clinical Practice, 2013, 99, e5-e8.	1.1	4
432	Neonatal diabetes in an infant of diabetic mother: same novel INS missense mutation in the mother and her offspring. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 745-8.	0.4	4

#	Article	IF	Citations
433	Three cases of Wolfram syndrome with different clinical aspects. Journal of Pediatric Endocrinology and Metabolism, 2014, 28, 433-8.	0.4	4
434	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
435	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
436	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
437	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
438	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. Endocrinology, Diabetes and Metabolism Case Reports, 2019, 2019, .	0.2	4
439	Permanent neonatal diabetes mellitus due to an ABCC8 mutation: a case report. JOP: Journal of the Pancreas, 2014, 15, 198-200.	1.5	4
440	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. Wellcome Open Research, 0, 2, 49.	0.9	4
441	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
442	A genetically engineered V79 cell line SD1 expressing rat CYP2B1 exhibits chromosomal instability at the integration site of the transfected DNA. Mutagenesis, 1995, 10, 549-554.	1.0	3
443	Multiple Endocrine Neoplasia Types 1 and 2. , 2004, 92, 267-284.		3
444	Neonatal diabetes is more than just a paediatric problem: 57 years of diabetes from a Kir6.2 mutation. Practical Diabetes International: the International Journal for Diabetes Care Teams Worldwide, 2005, 22, 342-344.	0.2	3
445	Gene duplications resulting in over expression of glucokinase are not a common cause of hypoglycaemia of infancy in humans. Molecular Genetics and Metabolism, 2008, 94, 268-269.	0.5	3
446	Genetic Testing in Diabetes Mellitus. , 2010, , 17-25.		3
447	Galactokinase Deficiency in a Patient with Congenital Hyperinsulinism. JIMD Reports, 2011, 5, 7-11.	0.7	3
448	Early-onset, severe lipoatrophy in a patient with permanent neonatal diabetes mellitus secondary to a recessive mutation in the INS gene. Pediatric Diabetes, 2012, 13, e26-e29.	1.2	3
449	Comment on: Khurana et al. The Diagnosis of Neonatal Diabetes in a Mother at 25 Years of Age. Diabetes Care 2012;35:e59. Diabetes Care, 2013, 36, e31-e31.	4.3	3
450	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

#	Article	IF	Citations
451	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
452	Diagnostic RET genetic testing in 1,058 index patients: A UK centre perspective. Clinical Endocrinology, 2021, 95, 295-302.	1.2	3
453	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
454	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	2.8	3
455	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	0.9	3
456	Successful discontinuation of insulin treatment after gestational diabetes is shown to be a case of MODY due to a glucokinase mutation. Open Medicine (Poland), 2008, 3, 225-228.	0.6	2
457	Coincidence of a Novel KCNJ11 Missense Variant R365H With a Paternally Inherited 6q24 Duplication in a Patient With Transient Neonatal Diabetes. Diabetes Care, 2008, 31, 1736-1737.	4.3	2
458	Evaluation of 13q14 Status in Multiple Myeloma by Digital Single Nucleotide Polymorphism Technology. Journal of Molecular Diagnostics, 2009, 11, 450-457.	1.2	2
459	Many patients have an identifiable genetic cause of Hirschsprung's disease. BMJ, The, 2012, 345, e8199-e8199.	3.0	2
460	Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 1065-9.	0.4	2
461	Next-Generation Sequencing for the Diagnosis of Monogenic Diabetes and Discovery of Novel Aetiologies. Frontiers in Diabetes, 2014, , 71-86.	0.4	2
462	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular Genetics & Genomic Medicine, 2021, , e1674.	0.6	2
463	Identification of Mutations in the Kir6.2 Subunit of the KATP Channel. Methods in Molecular Biology, 2008, 491, 235-245.	0.4	2
464	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, 0, , .	0.4	2
465	Congenital hyperinsulinism caused by mutations in ABCC8 (SUR1) gene. Indian Pediatrics, 2011, 48, 733-4.	0.2	2
466	Haemochromatosis and type 2 diabetes. Lancet, The, 1998, 352, 1068.	6.3	1
467	DWP6-4 Novel hepatocyte nuclear factor 4-alpha (HNF4A) promoter mutation in a New Zealand family. Diabetes Research and Clinical Practice, 2008, 79, S30.	1.1	1
468	Spondylocostal Dysostosis Associated with Methylmalonic Aciduria. Genetic Testing and Molecular Biomarkers, 2009, 13, 181-183.	0.3	1

#	Article	IF	Citations
469	A pathway to insulin independence in newborns and infants with diabetes. Journal of Perinatology, 2011, 31, 567-570.	0.9	1
470	Characterization of ABCC8 and KCNJ11 gene mutations and phenotypes in Korean patients with congenital hyperinsulinism. European Journal of Endocrinology, 2011, 165, 485-486.	1.9	1
471	Permanent neonatal diabetes due to a novel L105P (c.314T>C; p.Leu105Pro) heterozygous mutation in insulin gene. International Journal of Diabetes in Developing Countries, 2013, 33, 226-228.	0.3	1
472	Permanent neonatal diabetes misdiagnosed as type 1 diabetes in a 28-year-old female: A life-changing diagnosis. Diabetes Research and Clinical Practice, 2014, 106, e22-e24.	1.1	1
473	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
474	A novel autosomal recessive DEAF1 nonsense variant: expanding the clinical phenotype. Clinical Dysmorphology, 2020, 29, 114-117.	0.1	1
475	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
476	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. Wellcome Open Research, 2020, 5, 15.	0.9	1
477	Misannotation of multiple-nucleotide variants risks misdiagnosis. Wellcome Open Research, 2019, 4, 145.	0.9	1
478	Phenotype of a transient neonatal diabetes point mutation (SUR1-R1183W) in mice. Wellcome Open Research, 2020, 5, 15.	0.9	1
479	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
480	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
481	RET gene mutations are not a common cause of congenital solitary functioning kidney in adults. CKJ: Clinical Kidney Journal, 2009, 2, 183-184.	1.4	0
482	Genetic sequencing breakthrough to aid treatment for congenital hyperinsulinism. British Journal of Hospital Medicine (London, England: 2005), 2013, 74, 68-68.	0.2	0
483	Sirolimus therapy following subtotal pancreatectomy in neonatal hyperinsulinemic hypoglycaemia: a case report. International Journal of Pediatric Endocrinology (Springer), 2015, 2015, .	1.6	0
484	SP030HNF1B WHOLE-GENE DELETIONS ARE ASSOCIATED WITH AUTISTIC TRAITS. Nephrology Dialysis Transplantation, 2015, 30, iii390-iii390.	0.4	0
485	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
486	Insights from Monogenic Diabetes. , 2016, , 223-240.		0

#	Article	IF	Citations
487	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
488	Cover Image, Volume 176A, Number 9, September 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
489	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
490	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
491	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
492	ICR142 Benchmarker: evaluating, optimising and benchmarking variant calling performance using the ICR142 NGS validation series. Wellcome Open Research, 2018, 3, 108.	0.9	0
493	ATAD3 gene-familian de novo duplikazioek Harel-Yoon sindromea sortzen dute, eta kolesterolaren eta mitokondrien metabolismoan kalteak eragiten dituzte. , 0, , .		0
494	Title is missing!. , 2020, 15, e0228417.		0
495	Title is missing!. , 2020, 15, e0228417.		0
496	Title is missing!. , 2020, 15, e0228417.		0
497	Title is missing!. , 2020, 15, e0228417.		0