Kevin Colclough

List of Publications by Year in descending order

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41 papers

2,289 citations

304368 22 h-index 37 g-index

42 all docs 42 docs citations

42 times ranked 2439 citing authors

#	Article	IF	Citations
1	Update on mutations in glucokinase (<i>GCK</i>), which cause maturity-onset diabetes of the young, permanent neonatal diabetes, and hyperinsulinemic hypoglycemia. Human Mutation, 2009, 30, 1512-1526.	1.1	403
2	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
3	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
4	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
5	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
6	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
7	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
8	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. Nature Communications, 2017, 8, 888.	5.8	95
9	Monogenic diabetes: a gateway to precision medicine in diabetes. Journal of Clinical Investigation, 2021, 131, .	3.9	77
10	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
11	The Common p.R114W <i>HNF4A</i> Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. Diabetes, 2016, 65, 3212-3217.	0.3	46
12	Characteristics of maturity onset diabetes of the young in a large diabetes center. Pediatric Diabetes, 2016, 17, 360-367.	1.2	44
13	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
14	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
15	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.4	39
16	Polygenic Risk Variants for Type 2 Diabetes Susceptibility Modify Age at Diagnosis in Monogenic <i>HNF1A</i> Diabetes. Diabetes, 2010, 59, 266-271.	0.3	37
17	Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum Stress. Diabetes, 2021, 70, 1006-1018.	0.3	37
18	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

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19	Home urine C-peptide creatinine ratio testing can identify type 2 and MODY in pediatric diabetes. Pediatric Diabetes, 2012, 14, n/a-n/a.	1.2	29
20	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
21	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.	3.9	27
22	Evaluation of Evidence for Pathogenicity Demonstrates That <i>BLK</i> , <i>KLF11</i> , and <i>PAX4</i> Should Not Be Included in Diagnostic Testing for MODY. Diabetes, 2022, 71, 1128-1136.	0.3	27
23	Clinical utility gene card for: Maturity-onset diabetes of the young. European Journal of Human Genetics, 2014, 22, 1153-1153.	1.4	26
24	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
25	Diagnosis of monogenic diabetes: 10â€ Y ear experience in a large multiâ€ethnic diabetes center. Journal of Diabetes Investigation, 2016, 7, 332-337.	1.1	21
26	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.	1.8	19
27	Homozygous Hypomorphic <i>HNF1A</i> Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care, 2020, 43, 909-912.	4.3	13
28	Towards a systematic nationwide screening strategy for MODY. Diabetologia, 2017, 60, 609-612.	2.9	12
29	Evaluation of pregnancy outcomes in women with GCKâ€MODY. Diabetic Medicine, 2021, 38, e14488.	1.2	12
30	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. Diabetologia, 2022, 65, 336-342.	2.9	12
31	How do I diagnose Maturity Onset Diabetes of the Young in my patients?. Clinical Endocrinology, 2022, 97, 436-447.	1.2	11
32	Heterozygous Insulin Receptor (INSR) Mutation Associated with Neonatal Hyperinsulinemic Hypoglycaemia and Familial Diabetes Mellitus: Case Series. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2020, 12, 420-426.	0.4	8
33	Congenital hyperinsulinism due to mutations in HNF1A. European Journal of Medical Genetics, 2020, 63, 103928.	0.7	5
34	Case report: adult onset diabetes with partial pancreatic agenesis and congenital heart disease due to a de novo GATA6 mutation. BMC Medical Genetics, 2020, 21, 70.	2.1	4
35	Expanding the Phenotype of TRMT10A Mutations: Case Report and a Review of the Existing Cases. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2023, 15, 90-96.	0.4	3
36	Updated prevalence of monogenic diabetes in Australia: Fremantle Diabetes Study Phase 2. Medical Journal of Australia, 2019, 211, 189.	0.8	2

#	Article	lF	CITATIONS
37	Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. Diabetologia, 2022, 65, 246-249.	2.9	2
38	A novel heterozygous mutation in the insulin receptor gene presenting with type A severe insulin resistance syndrome. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 809-812.	0.4	1
39	Misannotation of multiple-nucleotide variants risks misdiagnosis. Wellcome Open Research, 2019, 4, 145.	0.9	1
40	Tests aiding diagnosis of monogenic diabetes. Practical Diabetes, 2017, 34, 217.	0.1	0
41	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;43:909–912. Diabetes Care, 2020, 43, e155-e156.	4.3	0