Kevin Colclough

List of Publications by Year
in descending order

41 papers	2,289 citations	304368 22 h-index	329751 37 g-index
42 all docs	42 docs citations	42 times ranked	2439 citing authors

#	Article	IF	Citations
1	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
2	Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. Diabetologia, 2022, 65, 246-249.	2.9	2
3	Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. Diabetologia, 2022, 65, 336-342.	2.9	12
4	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
5	Evaluation of Evidence for Pathogenicity Demonstrates That BLK, KLF11, and PAX4 Should Not Be Included in Diagnostic Testing for MODY. Diabetes, 2022, 71, 1128-1136.	0.3	27
6	How do I diagnose Maturity Onset Diabetes of the Young in my patients?. Clinical Endocrinology, 2022, 97, 436-447.	1.2	11
7	Evaluation of pregnancy outcomes in women with GCKâ€MODY. Diabetic Medicine, 2021, 38, e14488.	1.2	12
8	Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum Stress. Diabetes, 2021, 70, 1006-1018.	0.3	37
9	Monogenic diabetes: a gateway to precision medicine in diabetes. Journal of Clinical Investigation, 2021, 131, .	3.9	77
10	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
11	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
12	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
13	Congenital hyperinsulinism due to mutations in HNF1A. European Journal of Medical Genetics, 2020, 63, 103928.	0.7	5
14	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
15	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
16	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
17	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
18	Updated prevalence of monogenic diabetes in Australia: Fremantle Diabetes Study Phase 2. Medical Journal of Australia, 2019, 211, 189.	0.8	2

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19	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.4	39
20	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.	3.9	27
21	Misannotation of multiple-nucleotide variants risks misdiagnosis. Wellcome Open Research, 2019, 4, 145.	0.9	1
22	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.	1.8	19
23	Towards a systematic nationwide screening strategy for MODY. Diabetologia, 2017, 60, 609-612.	2.9	12
24	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
25	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. Nature Communications, 2017, 8, 888.	5.8	95
26	Tests aiding diagnosis of monogenic diabetes. Practical Diabetes, 2017, 34, 217.	0.1	0
27	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
28	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
29	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
30	The Common p.R114W HNF4A Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. Diabetes, 2016, 65, 3212-3217.	0.3	46
31	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
32	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
33	Characteristics of maturity onset diabetes of the young in a large diabetes center. Pediatric Diabetes, 2016, 17, 360-367.	1.2	44
34	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
35	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
36	Clinical utility gene card for: Maturity-onset diabetes of the young. European Journal of Human Genetics, 2014, 22, 1153-1153.	1.4	26

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37	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
38	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
39	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
40	Polygenic Risk Variants for Type 2 Diabetes Susceptibility Modify Age at Diagnosis in Monogenic HNF1A Diabetes. Diabetes, 2010, 59, 266-271.	0.3	37
41	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