Kashyap A Patel

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/6817167/publications.pdf

Version: 2024-02-01

55 papers	2,612 citations	304602 22 h-index	214721 47 g-index
63 all docs	63 docs citations	63 times ranked	4751 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	MANF supports the inner hair cell synapse and the outer hair cell stereocilia bundle in the cochlea. Life Science Alliance, 2022, 5, e202101068.	1.3	3
5	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
6	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
7	SavvyCNV: Genome-wide CNV calling from off-targetÂreads. PLoS Computational Biology, 2022, 18, e1009940.	1.5	18
8	PLIN1 Haploinsufficiency Causes a Favorable Metabolic Profile. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2318-e2323.	1.8	7
9	Atypical comorbidities in a child considered to have type 1 diabetes led to the diagnosis of SLC29A3 spectrum disorder. Hormones, 2022, 21, 501-506.	0.9	2
10	How do I diagnose Maturity Onset Diabetes of the Young in my patients?. Clinical Endocrinology, 2022, 97, 436-447.	1.2	11
11	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
12	Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients With Sulfonylurea-Treated ABCC8 Permanent Neonatal Diabetes. Diabetes Care, 2021, 44, 35-42.	4.3	24
13	Type 2 Diabetes and COVID-19â€"Related Mortality in the Critical Care Setting: A National Cohort Study in England, Marchâ€"July 2020. Diabetes Care, 2021, 44, 50-57.	4.3	139
14	More on STAT1 Gain of Function, Type 1 Diabetes, and JAK Inhibition. New England Journal of Medicine, 2021, 384, 93-94.	13.9	2
15	Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum Stress. Diabetes, 2021, 70, 1006-1018.	0.3	37
16	Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example from a recent study of MC4R. Cell, 2021, 184, 1651.	13.5	8
17	DR15-DQ6 remains dominantly protective against type 1 diabetes throughout the first five decades of life. Diabetologia, 2021, 64, 2258-2265.	2.9	8
18	Birth weight and diazoxide unresponsiveness strongly predict the likelihood of congenital hyperinsulinism due to a mutation in ABCC8 or KCNJ11. European Journal of Endocrinology, 2021, 185, 813-818.	1.9	2

#	Article	IF	Citations
19	Transient Neonatal Diabetes: An Etiologic Clue for the Adult Diabetologist. Canadian Journal of Diabetes, 2020, 44, 128-130.	0.4	5
20	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
21	Type 1 diabetes genetic risk score is discriminative of diabetes in non-Europeans: evidence from a study in India. Scientific Reports, 2020, 10, 9450.	1.6	25
22	Studies of insulin and proinsulin in pancreas and serum support the existence of aetiopathological endotypes of type 1 diabetes associated with age at diagnosis. Diabetologia, 2020, 63, 1258-1267.	2.9	98
23	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. Wellcome Open Research, 2020, 5, 175.	0.9	2
24	YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress. Journal of Clinical Investigation, 2020, 130, 6338-6353.	3.9	58
25	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. Wellcome Open Research, 2020, 5, 175.	0.9	1
26	Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a Population Setting. American Journal of Human Genetics, 2019, 104, 275-286.	2.6	158
27	Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated. Diabetes, 2019, 68, 1528-1535.	0.3	22
28	Patterns of postmeal insulin secretion in individuals with sulfonylurea-treated KCNJ11 neonatal diabetes show predominance of non-KATP-channel pathways. BMJ Open Diabetes Research and Care, 2019, 7, e000721.	1.2	9
29	Zinc Transporter 8 Autoantibodies (ZnT8A) and a Type 1 Diabetes Genetic Risk Score Can Exclude Individuals With Type 1 Diabetes From Inappropriate Genetic Testing for Monogenic Diabetes. Diabetes Care, 2019, 42, e16-e17.	4.3	19
30	Utility of systematic TSHR gene testing in adults with hyperthyroidism lacking overt autoimmunity and diffuse uptake on thyroid scintigraphy. Clinical Endocrinology, 2019, 90, 328-333.	1.2	4
31	Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.	3.9	27
32	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
33	Metformin selectively targets redox control of complex I energy transduction. Redox Biology, 2018, 14, 187-197.	3.9	115
34	PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.	1.8	19
35	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
36	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

#	Article	IF	Citations
37	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
38	Hyperglycaemiaâ€related complications at the time of diagnosis can cause permanent neurological disability in children with neonatal diabetes. Diabetic Medicine, 2017, 34, 1000-1004.	1.2	8
39	Precision diabetes: learning from monogenic diabetes. Diabetologia, 2017, 60, 769-777.	2.9	237
40	Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance. Nature Communications, 2017, 8, 888.	5.8	95
41	Interpretation of thyroid scintigraphy is inconsistent among endocrinologists. Journal of Endocrinological Investigation, 2017, 40, 1155-1157.	1.8	3
42	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
43	Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes. Diabetes, 2016, 65, 2094-2099.	0.3	146
44	Differential regulation of serum microRNA expression by HNF1Î² and HNF1Î± transcription factors. Diabetologia, 2016, 59, 1463-1473.	2.9	18
45	Investigation of salicylate hepatic responses in comparison with chemical analogues of the drug. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1412-1422.	1.8	8
46	The Common p.R114W HNF4A Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes. Diabetes, 2016, 65, 3212-3217.	0.3	46
47	Prospective functional classification of all possible missense variants in PPARG. Nature Genetics, 2016, 48, 1570-1575.	9.4	210
48	A Type 1 Diabetes Genetic Risk Score Can Aid Discrimination Between Type 1 and Type 2 Diabetes in Young Adults. Diabetes Care, 2016, 39, 337-344.	4.3	231
49	The LKB1-salt-inducible kinase pathway functions as a key gluconeogenic suppressor in the liver. Nature Communications, 2014, 5, 4535.	5.8	131
50	Glucose-6-Phosphateâ€“Mediated Activation of Liver Glycogen Synthase Plays a Key Role in Hepatic Glycogen Synthesis. Diabetes, 2013, 62, 4070-4082.	0.3	78
51	Cellular Responses to the Metal-Binding Properties of Metformin. Diabetes, 2012, 61, 1423-1433.	0.3	85
52	The AMPK-related kinase SIK2 is regulated by cAMP via phosphorylation at Ser358 in adipocytes. Biochemical Journal, 2012, 444, 503-514.	1.7	60
53	Thyroid disease in pregnancy. The Obstetrician and Gynaecologist, 2009, 11, 150-151.	0.2	0
54	The Absence of Islet Autoantibodies in Clinically Diagnosed Older-Adult Onset Type 1 Diabetes Suggests an Alternative Pathology, Advocating for Routine Testing in This Age Group. SSRN Electronic Journal, 0, , .	0.4	1

#	Article	IF	Citations
55	All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes identify women with a higher genetic risk for type 2 diabetes. Wellcome Open Research, 0, 5, 175.	0.9	0