## Kashyap A Patel

List of Publications by Year in descending order

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Κλομγλό Δ Όλτει

| #  | Article  | IF  | CITATIONS |
|----|--|-----|-----------|
| 1  | Precision diabetes: learning from monogenic diabetes. Diabetologia, 2017, 60, 769-777.   | 2.9 | 237       |
| 2  | A Type 1 Diabetes Genetic Risk Score Can Aid Discrimination Between Type 1 and Type 2 Diabetes in Young<br>Adults. Diabetes Care, 2016, 39, 337-344.   | 4.3 | 231       |
| 3  | Prospective functional classification of all possible missense variants in PPARG. Nature Genetics, 2016, 48, 1570-1575.  | 9.4 | 210       |
| 4  | Assessing the Pathogenicity, Penetrance, and Expressivity of Putative Disease-Causing Variants in a<br>Population Setting. American Journal of Human Genetics, 2019, 104, 275-286.   | 2.6 | 158       |
| 5  | Type 1 Diabetes Genetic Risk Score: A Novel Tool to Discriminate Monogenic and Type 1 Diabetes.<br>Diabetes, 2016, 65, 2094-2099.  | 0.3 | 146       |
| 6  | Type 2 Diabetes and COVID-19–Related Mortality in the Critical Care Setting: A National Cohort Study<br>in England, March–July 2020. Diabetes Care, 2021, 44, 50-57.   | 4.3 | 139       |
| 7  | The LKB1-salt-inducible kinase pathway functions as a key gluconeogenic suppressor in the liver.<br>Nature Communications, 2014, 5, 4535.  | 5.8 | 131       |
| 8  | Effectiveness and safety of long-term treatment with sulfonylureas in patients with neonatal diabetes<br>due to KCNJ11 mutations: an international cohort study. Lancet Diabetes and Endocrinology,the, 2018,<br>6, 637-646. | 5.5 | 120       |
| 9  | Metformin selectively targets redox control of complex I energy transduction. Redox Biology, 2018, 14, 187-197.  | 3.9 | 115       |
| 10 | 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        |
| 11 | Heterozygous RFX6 protein truncating variants are associated with MODY with reduced penetrance.<br>Nature Communications, 2017, 8, 888.  | 5.8 | 95        |
| 12 | Cellular Responses to the Metal-Binding Properties of Metformin. Diabetes, 2012, 61, 1423-1433.  | 0.3 | 85        |
| 13 | Glucose-6-Phosphate–Mediated Activation of Liver Glycogen Synthase Plays a Key Role in Hepatic<br>Glycogen Synthesis. Diabetes, 2013, 62, 4070-4082.   | 0.3 | 78        |
| 14 | 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        |
| 15 | 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        |
| 16 | The AMPK-related kinase SIK2 is regulated by cAMP via phosphorylation at Ser358 in adipocytes.<br>Biochemical Journal, 2012, 444, 503-514.   | 1.7 | 60        |
| 17 | YIPF5 mutations cause neonatal diabetes and microcephaly through endoplasmic reticulum stress.<br>Journal of Clinical Investigation, 2020, 130, 6338-6353.   | 3.9 | 58        |
| 18 | The Common p.R114W <i>HNF4A</i> Mutation Causes a Distinct Clinical Subtype of Monogenic Diabetes.<br>Diabetes, 2016, 65, 3212-3217.   | 0.3 | 46        |

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|----|--|-----|-----------|
| 19 | Loss of MANF Causes Childhood-Onset Syndromic Diabetes Due to Increased Endoplasmic Reticulum<br>Stress. Diabetes, 2021, 70, 1006-1018.  | 0.3 | 37        |
| 20 | 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        |
| 21 | 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        |
| 22 | Prediction algorithms: pitfalls in interpreting genetic variants of autosomal dominant monogenic diabetes. Journal of Clinical Investigation, 2019, 130, 14-16.  | 3.9 | 27        |
| 23 | Evaluation of Evidence for Pathogenicity Demonstrates That <i>BLK</i> , <i>KLF11</i> , and <i>PAX4</i><br>Should Not Be Included in Diagnostic Testing for MODY. Diabetes, 2022, 71, 1128-1136.                                    | 0.3 | 27        |
| 24 | Type 1 diabetes genetic risk score is discriminative of diabetes in non-Europeans: evidence from a study<br>in India. Scientific Reports, 2020, 10, 9450.  | 1.6 | 25        |
| 25 | Type 1 diabetes can present before the age of 6Âmonths and is characterised by autoimmunity and rapid<br>loss of beta cells. Diabetologia, 2020, 63, 2605-2615.  | 2.9 | 24        |
| 26 | Long-term Follow-up of Glycemic and Neurological Outcomes in an International Series of Patients<br>With Sulfonylurea-Treated <i>ABCC8</i> Permanent Neonatal Diabetes. Diabetes Care, 2021, 44, 35-42.                            | 4.3 | 24        |
| 27 | Trisomy 21 Is a Cause of Permanent Neonatal Diabetes That Is Autoimmune but Not HLA Associated.<br>Diabetes, 2019, 68, 1528-1535.  | 0.3 | 22        |
| 28 | PLIN1 Haploinsufficiency Is Not Associated With Lipodystrophy. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 3225-3230.   | 1.8 | 19        |
| 29 | Zinc Transporter 8 Autoantibodies (ZnT8A) and a Type 1 Diabetes Genetic Risk Score Can Exclude<br>Individuals With Type 1 Diabetes From Inappropriate Genetic Testing for Monogenic Diabetes. Diabetes<br>Care, 2019, 42, e16-e17. | 4.3 | 19        |
| 30 | Differential regulation of serum microRNA expression by HNF1β and HNF1α transcription factors.<br>Diabetologia, 2016, 59, 1463-1473.   | 2.9 | 18        |
| 31 | SavvyCNV: Genome-wide CNV calling from off-targetÂreads. PLoS Computational Biology, 2022, 18, e1009940.   | 1.5 | 18        |
| 32 | The Common <i>HNF1A</i> Variant I27L Is a Modifier of Age at Diabetes Diagnosis in Individuals With HNF1A-MODY. Diabetes, 2018, 67, 1903-1907.   | 0.3 | 12        |
| 33 | Systematic genetic testing for recessively inherited monogenic diabetes: a cross-sectional study in paediatric diabetes clinics. Diabetologia, 2022, 65, 336-342.  | 2.9 | 12        |
| 34 | How do I diagnose Maturity Onset Diabetes of the Young in my patients?. Clinical Endocrinology, 2022, 97, 436-447.   | 1.2 | 11        |
| 35 | Patterns of postmeal insulin secretion in individuals with sulfonylurea-treated KCNJ11 neonatal<br>diabetes show predominance of non-KATP-channel pathways. BMJ Open Diabetes Research and Care,<br>2019, 7, e000721.              | 1.2 | 9         |
| 36 | Investigation of salicylate hepatic responses in comparison with chemical analogues of the drug.<br>Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1412-1422.   | 1.8 | 8         |

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|----|--|------|-----------|
| 37 | Hyperglycaemiaâ€related complications at the time of diagnosis can cause permanent neurological<br>disability in children with neonatal diabetes. Diabetic Medicine, 2017, 34, 1000-1004.                                | 1.2  | 8         |
| 38 | Unreliability of genotyping arrays for detecting very rare variants in human genetic studies: Example<br>from a recent study of MC4R. Cell, 2021, 184, 1651.   | 13.5 | 8         |
| 39 | DR15-DQ6 remains dominantly protective against type 1 diabetes throughout the first five decades of life. Diabetologia, 2021, 64, 2258-2265.   | 2.9  | 8         |
| 40 | <i>PLIN1</i> Haploinsufficiency Causes a Favorable Metabolic Profile. Journal of Clinical<br>Endocrinology and Metabolism, 2022, 107, e2318-e2323.   | 1.8  | 7         |
| 41 | Transient Neonatal Diabetes: An Etiologic Clue for the Adult Diabetologist. Canadian Journal of<br>Diabetes, 2020, 44, 128-130.  | 0.4  | 5         |
| 42 | Utility of systematic <i>TSHR</i> gene testing in adults with hyperthyroidism lacking overt<br>autoimmunity and diffuse uptake on thyroid scintigraphy. Clinical Endocrinology, 2019, 90, 328-333.                       | 1.2  | 4         |
| 43 | Interpretation of thyroid scintigraphy is inconsistent among endocrinologists. Journal of Endocrinological Investigation, 2017, 40, 1155-1157.   | 1.8  | 3         |
| 44 | Expanding the Phenotype of TRMT10A Mutations: Case Report and a Review of the Existing Cases. JCRPE<br>Journal of Clinical Research in Pediatric Endocrinology, 2023, 15, 90-96.   | 0.4  | 3         |
| 45 | MANF supports the inner hair cell synapse and the outer hair cell stereocilia bundle in the cochlea.<br>Life Science Alliance, 2022, 5, e202101068.  | 1.3  | 3         |
| 46 | More on STAT1 Gain of Function, Type 1 Diabetes, and JAK Inhibition. New England Journal of Medicine, 2021, 384, 93-94.  | 13.9 | 2         |
| 47 | All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes<br>identify women with a higher genetic risk for type 2 diabetes. Wellcome Open Research, 2020, 5, 175.                    | 0.9  | 2         |
| 48 | Association of birthweight and penetrance of diabetes in individuals with HNF4A-MODY: a cohort study. Diabetologia, 2022, 65, 246-249.   | 2.9  | 2         |
| 49 | Birth weight and diazoxide unresponsiveness strongly predict the likelihood of congenital<br>hyperinsulinism due to a mutation in ABCC8 or KCNJ11. European Journal of Endocrinology, 2021, 185,<br>813-818.             | 1.9  | 2         |
| 50 | 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         |
| 51 | The Absence of Islet Autoantibodies in Clinically Diagnosed Older-Adult Onset Type 1 Diabetes Suggests<br>an Alternative Pathology, Advocating for Routine Testing in This Age Group. SSRN Electronic Journal,<br>0, , . | 0.4  | 1         |
| 52 | All thresholds of maternal hyperglycaemia from the WHO 2013 criteria for gestational diabetes<br>identify women with a higher genetic risk for type 2 diabetes. Wellcome Open Research, 2020, 5, 175.                    | 0.9  | 1         |
| 53 | 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         |
| 54 | Thyroid disease in pregnancy. The Obstetrician and Gynaecologist, 2009, 11, 150-151.   | 0.2  | 0         |

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