Pratik Shah

List of Publications by Citations

55 papers	656 citations	15 h-index	24 g-index
61 ext. papers	854 ext. citations	5 avg, IF	3.99 L-index

#	Paper	IF	Citations
55	Sirolimus therapy in infants with severe hyperinsulinemic hypoglycemia. *New England Journal of Medicine*, **2014**, 370, 1131-7	59.2	97
54	Hyperinsulinaemic hypoglycaemia in children and adults. *Lancet Diabetes and Endocrinology,the*, **2017**, 5, 729-742	18.1	52
53	Therapies and outcomes of congenital hyperinsulinism-induced hypoglycaemia. *Diabetic Medicine*, **2019**, 36, 9-21	3.5	48
52	The Diagnosis and Management of Hyperinsulinaemic Hypoglycaemia. *JCRPE Journal of Clinical Research in Pediatric Endocrinology*, **2015**, 7, 86-97	1.9	44
51	Long-term follow-up of children with congenital hyperinsulinism on octreotide therapy. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, 3660-7	5.6	42
50	Persistent hyperinsulinaemic hypoglycaemia in infancy. *Seminars in Pediatric Surgery*, **2014**, 23, 76-82	2.1	28
49	Pigmentary hypertrichosis and non-autoimmune insulin-dependent diabetes mellitus (PHID) syndrome is associated with severe chronic inflammation and cardiomyopathy, and represents a new monogenic autoinflammatory syndrome. *Journal of Pediatric Endocrinology and Metabolism*, **2013**, 26, 877-82	1.6	27
48	Clinical and histological heterogeneity of congenital hyperinsulinism due to paternally inherited heterozygous ABCC8/KCNJ11 mutations. *European Journal of Endocrinology*, **2014**, 171, 685-95	6.5	27
47	Impaired EIF2S3 function associated with a novel phenotype of X-linked hypopituitarism with glucose dysregulation. *EBioMedicine*, **2019**, 42, 470-480	8.8	24
46	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 9 affected individuals. *Genetics in Medicine*, **2019**, 21, 233-242	8.1	23
45	Hepatocyte Nuclear Factor-4 Alfa Mutation Associated with Hyperinsulinaemic Hypoglycaemia and Atypical Renal Fanconi Syndrome: Expanding the Clinical Phenotype. *Hormone Research in Paediatrics*, **2016**, 86, 337-341	3.3	21
44	Use of Long-Acting Somatostatin Analogue (Lanreotide) in an Adolescent with Diazoxide-Responsive Congenital Hyperinsulinism and Its Psychological Impact. *Hormone Research in Paediatrics*, **2015**, 84, 355-60	3.3	20
43	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	1.6	19
42	Hyperinsulinemic hypoglycemia in children and adolescents: Recent advances in understanding of pathophysiology and management. *Reviews in Endocrine and Metabolic Disorders*, **2020**, 21, 577-597	10.5	19
41	Assessment of Nifedipine Therapy in Hyperinsulinemic Hypoglycemia due to Mutations in the ABCC8 Gene. *Journal of Clinical Endocrinology and Metabolism*, **2017**, 102, 822-830	5.6	15
40	Sirolimus: Efficacy and Complications in Children With Hyperinsulinemic Hypoglycemia: A 5-Year Follow-Up Study. *Journal of the Endocrine Society*, **2019**, 3, 699-713	0.4	12
39	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	3.7	11

38	Diazoxide-induced pulmonary hypertension in hyperinsulinaemic hypoglycaemia: Recommendations from a multicentre study in the United Kingdom. *Clinical Endocrinology*, **2019**, 91, 770-775	3.4	10
37	Familial isolated growth hormone deficiency due to a novel homozygous missense mutation in the growth hormone releasing hormone receptor gene: clinical presentation with hypoglycemia. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E2730-4	5.6	10
36	Hyperinsulinaemic hypoglycaemia-an overview of a complex clinical condition. *European Journal of Pediatrics*, **2019**, 178, 1151-1160	4.1	9
35	Severe Hyperinsulinaemic Hypoglycaemia in Beckwith-Wiedemann Syndrome due to Paternal Uniparental Disomy of 11p15.5 Managed with Sirolimus Therapy. *Hormone Research in Paediatrics*, **2016**, 85, 353-7	3.3	8
34	The Use of a Long-Acting Somatostatin Analogue (Lanreotide) in Three Children with Focal Forms of Congenital Hyperinsulinaemic Hypoglycaemia. *Hormone Research in Paediatrics*, **2019**, 91, 56-61	3.3	7
33	Genetic Analysis of Pediatric Primary Adrenal Insufficiency of Unknown Etiology: 25 YearsT Experience in the UK. *Journal of the Endocrine Society*, **2021**, 5, bvab086	0.4	7
32	Hyperinsulinaemic hypoglycaemia: A new presentation of 16p11.2 deletion syndrome. *Clinical Endocrinology*, **2019**, 90, 766-769	3.4	7
31	Postprandial hyperinsulinaemic hypoglycaemia secondary to a congenital portosystemic shunt. *Hormone Research in Paediatrics*, **2015**, 83, 217-20	3.3	6
30	Ga-NODAGA-exendin-4 PET improves the detection of focal congenital hyperinsulinism. *Journal of Nuclear Medicine*, **2021**,	8.9	6
29	Mutations in MAGEL2 and L1CAM Are Associated With Congenital Hypopituitarism and Arthrogryposis. *Journal of Clinical Endocrinology and Metabolism*, **2019**, 104, 5737-5750	5.6	5
28	Sirolimus precipitating diabetes mellitus in a patient with congenital hyperinsulinaemic hypoglycaemia due to autosomal dominant ABCC8 mutation. *Journal of Pediatric Endocrinology and Metabolism*, **2017**, 30, 1219-1222	1.6	5
27	Persistent hyperinsulinaemic hypoglycaemia in children with Rubinstein-Taybi syndrome. *European Journal of Endocrinology*, **2019**, 181, 121-128	6.5	5
26	COVID-19 in Children and Adolescents with Endocrine Conditions. *Hormone and Metabolic Research*, **2020**, 52, 769-774	3.1	5
25	Towards enhanced understanding of idiopathic ketotic hypoglycemia: a literature review and introduction of the patient organization, Ketotic Hypoglycemia International. *Orphanet Journal of Rare Diseases*, **2021**, 16, 173	4.2	5
24	Gastrointestinal dysmotility and pancreatic insufficiency in 2 siblings with Donohue syndrome. *Pediatric Diabetes*, **2017**, 18, 839-843	3.6	4
23	Post-Prandial Hyperinsulinaemic Hypoglycaemia after Oesophageal Surgery in Children. *Hormone Research in Paediatrics*, **2019**, 91, 216-220	3.3	3
22	Genetic Characterization of Short Stature Patients With Overlapping Features of Growth Hormone Insensitivity Syndromes. *Journal of Clinical Endocrinology and Metabolism*, **2021**, 106, e4716-e4733	5.6	3
21	Cenani-Lenz syndactyly in siblings with a novel homozygous LRP4 mutation and recurrent hypoglycaemia. *Clinical Dysmorphology*, **2020**, 29, 73-80	0.9	2

20	Cushing syndrome in a child due to pro-opiomelanocortin (POMC) secretion from a yolk sac tumor. *European Journal of Endocrinology*, **2017**, 176, K1-K7	6.5	2
19	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. *Endocrinology, Diabetes and Metabolism Case Reports*, **2019**, 2019,	1.4	2
18	Syndromic Forms of Hyperinsulinaemic Hypoglycaemia-A 15-year follow-up Study. *Clinical Endocrinology*, **2021**, 94, 399-412	3.4	2
17	Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 1065-9	1.6	1
16	Biallelic DNAJC3 variants in a neuroendocrine developmental disorder with insulin dysregulation. *Clinical Dysmorphology*, **2022**, 31, 11-17	0.9	1
15	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
14	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. *Wellcome Open Research*, **2019**, 4, 149	4.8	1
13	Unusual Glycemic Presentations in a Child with a Novel Heterozygous Intragenic INSR Deletion. *Hormone Research in Paediatrics*, **2020**, 93, 396-401	3.3	1
12	Diazoxide-responsive hyperinsulinaemic hypoglycaemia in tyrosinaemia type 1. *Endocrinology, Diabetes and Metabolism Case Reports*, **2021**, 2021,	1.4	1
11	Variation in Glycemic Outcomes in Focal Forms of Congenital Hyperinsulinism-The UK Perspective.. *Journal of the Endocrine Society*, **2022**, 6, bvac033	0.4	1
10	Laparoscopic resection of pancreatic neck lesion with Roux-en-Y pancreatico-jejunostomy. *Journal of Pediatric Surgery Case Reports*, **2019**, 40, 71-75	0.3	0
9	Sotos Syndrome Presenting with Neonatal Hyperinsulinaemic Hypoglycaemia, Extensive Thrombosis, and Multisystem Involvement. *Hormone Research in Paediatrics*, **2019**, 92, 64-70	3.3	0
8	Reply: Ga NODAGA-Exendin-4 PET Scanning for Focal Congenital Hyperinsulinism: Need for Replication.. *Journal of Nuclear Medicine*, **2022**, 63, 493-494	8.9	0
7	Bradycardia in minor trauma: don't be slow on the uptake!. *Emergency Medicine Journal*, **2007**, 24, e13	1.5	
6	Disorders Associated with Hypoglycaemia in Children **2019**, 671-699		
5	Enteroinsular hormones in two siblings with Donohue syndrome and complete leptin deficiency. *Pediatric Diabetes*, **2018**, 19, 675-679	3.6	
4	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 **2020**, 15, e0228417		
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2 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 **2020**, 15, e0228417

1 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 **2020**, 15, e0228417