

Pratik Shah

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Biallelic DNAJC3 variants in a neuroendocrine developmental disorder with insulin dysregulation. <i>Clinical Dysmorphology</i> , 2022 , 31, 11-17	0.9	1
54	Variation in Glycemic Outcomes in Focal Forms of Congenital Hyperinsulinism-The UK Perspective.. <i>Journal of the Endocrine Society</i> , 2022 , 6, bvac033	0.4	1
53	Reply: Ga NODAGA-Exendin-4 PET Scanning for Focal Congenital Hyperinsulinism: Need for Replication.. <i>Journal of Nuclear Medicine</i> , 2022 , 63, 493-494	8.9	0
52	Towards enhanced understanding of idiopathic ketotic hypoglycemia: a literature review and introduction of the patient organization, Ketotic Hypoglycemia International. <i>Orphanet Journal of Rare Diseases</i> , 2021 , 16, 173	4.2	5
51	Genetic Analysis of Pediatric Primary Adrenal Insufficiency of Unknown Etiology: 25 YearsT Experience in the UK. <i>Journal of the Endocrine Society</i> , 2021 , 5, bvab086	0.4	7
50	Genetic Characterization of Short Stature Patients With Overlapping Features of Growth Hormone Insensitivity Syndromes. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021 , 106, e4716-e4733	5.6	3
49	Ga-NODAGA-exendin-4 PET improves the detection of focal congenital hyperinsulinism. <i>Journal of Nuclear Medicine</i> , 2021 ,	8.9	6
48	Syndromic Forms of Hyperinsulinaemic Hypoglycaemia-A 15-year follow-up Study. <i>Clinical Endocrinology</i> , 2021 , 94, 399-412	3.4	2
47	Diazoxide-responsive hyperinsulinaemic hypoglycaemia in tyrosinaemia type 1. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2021 , 2021,	1.4	1
46	Hyperinsulinemic hypoglycemia in children and adolescents: Recent advances in understanding of pathophysiology and management. <i>Reviews in Endocrine and Metabolic Disorders</i> , 2020 , 21, 577-597	10.5	19
45	Cenani-Lenz syndactyly in siblings with a novel homozygous LRP4 mutation and recurrent hypoglycaemia. <i>Clinical Dysmorphology</i> , 2020 , 29, 73-80	0.9	2
44	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. <i>PLoS ONE</i> , 2020 , 15, e0228417	3.7	11
43	Unusual Glycemic Presentations in a Child with a Novel Heterozygous Intragenic INSR Deletion. <i>Hormone Research in Paediatrics</i> , 2020 , 93, 396-401	3.3	1
42	COVID-19 in Children and Adolescents with Endocrine Conditions. <i>Hormone and Metabolic Research</i> , 2020 , 52, 769-774	3.1	5
41	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		
40	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		
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 2020 , 15, e0228417		

38	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		
37	Laparoscopic resection of pancreatic neck lesion with Roux-en-Y pancreatico-jejunostomy. <i>Journal of Pediatric Surgery Case Reports</i> , 2019 , 40, 71-75	0.3	0
36	Impaired EIF2S3 function associated with a novel phenotype of X-linked hypopituitarism with glucose dysregulation. <i>EBioMedicine</i> , 2019 , 42, 470-480	8.8	24
35	Sotos Syndrome Presenting with Neonatal Hyperinsulinaemic Hypoglycaemia, Extensive Thrombosis, and Multisystem Involvement. <i>Hormone Research in Paediatrics</i> , 2019 , 92, 64-70	3.3	0
34	Sirolimus: Efficacy and Complications in Children With Hyperinsulinemic Hypoglycemia: A 5-Year Follow-Up Study. <i>Journal of the Endocrine Society</i> , 2019 , 3, 699-713	0.4	12
33	Congenital hyperinsulinism as the presenting feature of Kabuki syndrome: clinical and molecular characterization of 9 affected individuals. <i>Genetics in Medicine</i> , 2019 , 21, 233-242	8.1	23
32	The Use of a Long-Acting Somatostatin Analogue (Lanreotide) in Three Children with Focal Forms of Congenital Hyperinsulinaemic Hypoglycaemia. <i>Hormone Research in Paediatrics</i> , 2019 , 91, 56-61	3.3	7
31	Post-Prandial Hyperinsulinaemic Hypoglycaemia after Oesophageal Surgery in Children. <i>Hormone Research in Paediatrics</i> , 2019 , 91, 216-220	3.3	3
30	Hyperinsulinaemic hypoglycaemia-an overview of a complex clinical condition. <i>European Journal of Pediatrics</i> , 2019 , 178, 1151-1160	4.1	9
29	Mutations in MAGEL2 and L1CAM Are Associated With Congenital Hypopituitarism and Arthrogyposis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2019 , 104, 5737-5750	5.6	5
28	Diazoxide-induced pulmonary hypertension in hyperinsulinaemic hypoglycaemia: Recommendations from a multicentre study in the United Kingdom. <i>Clinical Endocrinology</i> , 2019 , 91, 770-775	3.4	10
27	Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2019 , 2019,	1.4	2
26	Persistent hyperinsulinaemic hypoglycaemia in children with Rubinstein-Taybi syndrome. <i>European Journal of Endocrinology</i> , 2019 , 181, 121-128	6.5	5
25	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019 , 4, 149	4.8	1
24	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019 , 4, 149	4.8	1
23	Hyperinsulinaemic hypoglycaemia: A new presentation of 16p11.2 deletion syndrome. <i>Clinical Endocrinology</i> , 2019 , 90, 766-769	3.4	7
22	Disorders Associated with Hypoglycaemia in Children 2019 , 671-699		
21	Therapies and outcomes of congenital hyperinsulinism-induced hypoglycaemia. <i>Diabetic Medicine</i> , 2019 , 36, 9-21	3.5	48

20	Enteroinsular hormones in two siblings with Donohue syndrome and complete leptin deficiency. <i>Pediatric Diabetes</i> , 2018 , 19, 675-679	3.6	
19	Hyperinsulinaemic hypoglycaemia in children and adults. <i>Lancet Diabetes and Endocrinology</i> , 2017 , 5, 729-742	18.1	52
18	Gastrointestinal dysmotility and pancreatic insufficiency in 2 siblings with Donohue syndrome. <i>Pediatric Diabetes</i> , 2017 , 18, 839-843	3.6	4
17	Cushing syndrome in a child due to pro-opiomelanocortin (POMC) secretion from a yolk sac tumor. <i>European Journal of Endocrinology</i> , 2017 , 176, K1-K7	6.5	2
16	Sirolimus precipitating diabetes mellitus in a patient with congenital hyperinsulinaemic hypoglycaemia due to autosomal dominant ABCC8 mutation. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2017 , 30, 1219-1222	1.6	5
15	Assessment of Nifedipine Therapy in Hyperinsulinemic Hypoglycemia due to Mutations in the ABCC8 Gene. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2017 , 102, 822-830	5.6	15
14	Severe Hyperinsulinaemic Hypoglycaemia in Beckwith-Wiedemann Syndrome due to Paternal Uniparental Disomy of 11p15.5 Managed with Sirolimus Therapy. <i>Hormone Research in Paediatrics</i> , 2016 , 85, 353-7	3.3	8
13	Hepatocyte Nuclear Factor-4 Alfa Mutation Associated with Hyperinsulinaemic Hypoglycaemia and Atypical Renal Fanconi Syndrome: Expanding the Clinical Phenotype. <i>Hormone Research in Paediatrics</i> , 2016 , 86, 337-341	3.3	21
12	The Diagnosis and Management of Hyperinsulinaemic Hypoglycaemia. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2015 , 7, 86-97	1.9	44
11	Sirolimus therapy in a patient with severe hyperinsulinaemic hypoglycaemia due to a compound heterozygous ABCC8 gene mutation. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2015 , 28, 695-9	1.6	19
10	Postprandial hyperinsulinaemic hypoglycaemia secondary to a congenital portosystemic shunt. <i>Hormone Research in Paediatrics</i> , 2015 , 83, 217-20	3.3	6
9	Use of Long-Acting Somatostatin Analogue (Lanreotide) in an Adolescent with Diazoxide-Responsive Congenital Hyperinsulinism and Its Psychological Impact. <i>Hormone Research in Paediatrics</i> , 2015 , 84, 355-60	3.3	20
8	Sirolimus therapy in infants with severe hyperinsulinemic hypoglycemia. <i>New England Journal of Medicine</i> , 2014 , 370, 1131-7	59.2	97
7	Persistent hyperinsulinaemic hypoglycaemia in infancy. <i>Seminars in Pediatric Surgery</i> , 2014 , 23, 76-82	2.1	28
6	Long-term follow-up of children with congenital hyperinsulinism on octreotide therapy. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014 , 99, 3660-7	5.6	42
5	Chromosome 6q24 transient neonatal diabetes mellitus and protein sensitive hyperinsulinaemic hypoglycaemia. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014 , 27, 1065-9	1.6	1
4	Clinical and histological heterogeneity of congenital hyperinsulinism due to paternally inherited heterozygous ABCC8/KCNJ11 mutations. <i>European Journal of Endocrinology</i> , 2014 , 171, 685-95	6.5	27
3	Familial isolated growth hormone deficiency due to a novel homozygous missense mutation in the growth hormone releasing hormone receptor gene: clinical presentation with hypoglycemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014 , 99, E2730-4	5.6	10

- 2 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
- 1 Bradycardia in minor trauma: don't be slow on the uptake!. *Emergency Medicine Journal*, **2007**, *24*, e13 1.5