Tohru Yorifuji

List of Publications by Year
in descending order

56 papers	1,082 citations	567281 15 h-index	434195 31 g-index
56 all docs	56 docs citations	56 times ranked	1380 citing authors

#	Article	IF	Citations
1	Congenital hyperinsulinism: current status and future perspectives. Annals of Pediatric Endocrinology and Metabolism, 2014, 19, 57.	2.3	140
2	The C42R Mutation in the Kir6.2 (KCNJ11) Gene as a Cause of Transient Neonatal Diabetes, Childhood Diabetes, or Later-Onset, Apparently Type 2 Diabetes Mellitus. Journal of Clinical Endocrinology and Metabolism, 2005, 90, 3174-3178.	3.6	111
3	Temple syndrome: comprehensive molecular and clinical findings in 32 Japanese patients. Genetics in Medicine, 2017, 19, 1356-1366.	2.4	96
4	Comprehensive molecular analysis of Japanese patients with pediatric-onset MODY-type diabetes mellitus. Pediatric Diabetes, 2012, 13, 26-32.	2.9	73
5	Safety Outcomes During Pediatric GH Therapy: Final Results From the Prospective GeNeSIS Observational Program. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 379-389.	3.6	51
6	Clinical practice guidelines for congenital hyperinsulinism. Clinical Pediatric Endocrinology, 2017, 26, 127-152.	0.8	48
7	Frequencies of spontaneous breast development and spontaneous menarche in Turner syndrome in Japan. Clinical Pediatric Endocrinology, 2015, 24, 167-173.	0.8	43
8	Nationwide survey of endogenous hyperinsulinemic hypoglycemia in Japan (2017â€“2018): Congenital hyperinsulinism, insulinoma, nonâ€insulinoma pancreatogenous hypoglycemia syndrome and insulin autoimmune syndrome (Hirataâ€™s disease). Journal of Diabetes Investigation, 2020, 11, 554-563.	2.4	36
9	Molecular and Clinical Analysis of Japanese Patients with Persistent Congenital Hyperinsulinism: Predominance of Paternally Inherited Monoallelic Mutations in the KATPChannel Genes. Journal of Clinical Endocrinology and Metabolism, 2011, 96, E141-E145.	3.6	35
10	Dominantly inherited diabetes mellitus caused by GATA6 haploinsufficiency: variable intrafamilial presentation. Journal of Medical Genetics, 2012, 49, 642-643.	3.2	35
11	Efficacy and safety of longâ€term, continuous subcutaneous octreotide infusion for patients with different subtypes of K_{ATP}â€channel hyperinsulinism. Clinical Endocrinology, 2013, 78, 891-897.	2.4	35
12	Molecular and clinical features of K_{ATP} -channel neonatal diabetes mellitus in Japan. Pediatric Diabetes, 2017, 18, 532-539.	2.9	27
13	Efficacy and safety of octreotide for the treatment of congenital hyperinsulinism: a prospective, open-label clinical trial and an observational study in Japan using a nationwide registry. Endocrine Journal, 2017, 64, 867-880.	1.6	25
14	Clinical features and management of organic acidemias in Japan. Journal of Human Genetics, 2013, 58, 769-774.	2.3	24
15	Clinical characteristics of insulin resistance syndromes: A nationwide survey in Japan. Journal of Diabetes Investigation, 2020, 11, 603-616.	2.4	20
16	Relapsing 6q24-related transient neonatal diabetes mellitus successfully treated with a dipeptidyl peptidase-4 inhibitor: a case report. Pediatric Diabetes, 2014, 15, 606-610.	2.9	16
17	Genetic basis of early-onset, maturity-onset diabetes of the young-like diabetes in Japan and features of patients without mutations in the major MODY genes: Dominance of maternal inheritance. Pediatric Diabetes, 2018, 19, 1164-1172.	2.9	16
18	Lasting ¹⁸F-DOPA PET Uptake after Clinical Remission of the Focal Form of Congenital Hyperinsulinism. Hormone Research in Paediatrics, 2011, 76, 286-290.	1.8	15

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19	Chromosome 6q24-related diabetes mellitus. Clinical Pediatric Endocrinology, 2018, 27, 59-65.	0.8	15
20	Two patients with HNF4A-related congenital hyperinsulinism and renal tubular dysfunction: A clinical variation which includes transient hepatic dysfunction. Diabetes Research and Clinical Practice, 2015, 108, e53-e55.	2.8	14
21	Gonadal function, fertility, and reproductive medicine in childhood and adolescent cancer patients: a national survey of Japanese pediatric endocrinologists. Clinical Pediatric Endocrinology, 2016, 25, 45-57.	0.8	13
22	Circulating tricarboxylic acid cycle metabolite levels in citrin-deficient children with metabolic adaptation, with and without sodium pyruvate treatment. Molecular Genetics and Metabolism, 2017, 120, 207-212.	1.1	12
23	Heterogeneous nature of diabetes in a family with a gain-of-function mutation in the ATP-binding cassette subfamily C member 8 (ABCC8) gene. Endocrine Journal, 2018, 65, 1055-1059.	1.6	12
24	Incidence rate and characteristics of symptomatic vitamin D deficiency in children: a nationwide survey in Japan. Endocrine Journal, 2018, 65, 593-599.	1.6	12
25	Pregnancy outcome of Japanese patients with glucokinaseâ€“maturityâ€onset diabetes of the young. Journal of Diabetes Investigation, 2019, 10, 1586-1589.	2.4	12
26	Congenital hyperinsulinism: Global and <scp>J</scp>apanese perspectives. Pediatrics International, 2014, 56, 467-476.	0.5	11
27	Serum Amino Acid Profiling in Citrin-Deficient Children Exhibiting Normal Liver Function During the Apparently Healthy Period. JIMD Reports, 2018, 43, 53-61.	1.5	9
28	DEND syndrome due to V59A mutation in KCNJ11 gene: unresponsive to sulfonylureas. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 143-6.	0.9	8
29	Diagnosis of congenital hyperinsulinism: Biochemical profiles during hypoglycemia. Pediatric Diabetes, 2018, 19, 259-264.	2.9	8
30	Identification of a variant associated with earlyâ€onset diabetes in the intron of the insulin gene with exome sequencing. Journal of Diabetes Investigation, 2019, 10, 947-950.	2.4	8
31	Mutational and clinical spectrum of Japanese patients with hereditary hemorrhagic telangiectasia. BMC Medical Genomics, 2021, 14, 288.	1.5	8
32	Growth hormone activates hepatic and cerebral cholesterol metabolism in small-for-gestational age children without catch-up growth. Journal of Clinical Lipidology, 2017, 11, 1032-1042.	1.5	7
33	Successful treatment of infantile-onset ACAD9-related cardiomyopathy with a combination of sodium pyruvate, beta-blocker, and coenzyme Q₁₀. Journal of Pediatric Endocrinology and Metabolism, 2019, 32, 1181-1185.	0.9	7
34	Efficacy and Safety of Once-Weekly Somatrogon Compared with Once-Daily Somatropin (GenotropinÂ®) in Japanese Children with Pediatric Growth Hormone Deficiency: Results from a Randomized Phase 3 Study. Hormone Research in Paediatrics, 2022, 95, 275-285.	1.8	7
35	Changes of lipoproteins in phenylalanine hydroxylase-deficient children during the first year of life. Clinica Chimica Acta, 2014, 433, 1-4.	1.1	6
36	Identification of clinical factors related to antibodyâ€mediated immune response to the subfornical organ. Clinical Endocrinology, 2022, 97, 72-80.	2.4	6

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37	A case of CHARGE syndrome associated with hyperinsulinemic hypoglycemia in infancy. European Journal of Medical Genetics, 2018, 61, 312-314.	1.3	5
38	Cholesterol Metabolism Is Enhanced in the Liver and Brain of Children With Citrin Deficiency. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2488-2497.	3.6	5
39	Practice guideline for lipodystrophy syndromesâ€"clinically important diseases of the Japan Endocrine Society (JES). Endocrine Journal, 2021, 68, 1027-1042.	1.6	5
40	New classification and diagnostic criteria for insulin resistance syndrome. Endocrine Journal, 2022, 69, 107-113.	1.6	5
41	New classification and diagnostic criteria for insulin resistance syndrome. Diabetology International, 2022, 13, 337-343.	1.4	5
42	Growth Hormone Treatment for Achondroplasia. Pediatric Endocrinology Reviews, 2018, 16, 123-128.	1.2	5
43	Congenital hyperinsulinism treated by surgical resection of the hyperplastic lesion which had been preoperatively diagnosed by 18F-DOPA PET examination in Japan: a nationwide survey. Pediatric Surgery International, 2018, 34, 1093-1098.	1.4	4
44	Diabetes caused by Kir6.2 mutation: Successful treatment with oral glibenclamide switched from continuous subcutaneous insulin infusion in the early phase of the disease. Pediatrics International, 2012, 54, 277-279.	0.5	3
45	Focal form of congenital hyperinsulinism clearly detectable by contrast-enhanced computed tomography imaging. International Journal of Pediatric Endocrinology (Springer), 2015, 2015, 20.	1.6	3
46	Acute Myeloid Leukemia With RBM15-MKL1 Presenting as Severe Hepatic Failure. Global Pediatric Health, 2017, 4, 2333794X1668901.	0.7	3
47	Sulfonylurea treatment in an infant with transient neonatal diabetes mellitus caused by an adenosine triphosphate binding cassette subfamily C member 8 gene mutation. Clinical Pediatric Endocrinology, 2017, 26, 165-169.	0.8	3
48	Chromosome 6q24 methylation defects are uncommon in childhood-onset non-autoimmune diabetes mellitus patients born appropriate- or large-for-gestational age. Clinical Pediatric Endocrinology, 2016, 25, 99-102.	0.8	3
49	Case Report: A Difficult-to-Diagnose Case of Hyperinsulinemic Hypoglycemia Surgically Treated After Developing Acute Pancreatitis. Frontiers in Endocrinology, 2021, 12, 731071.	3.5	3
50	ApoE4 Determines the Reduction in LDL-C After GH Replacement Therapy in Children With an Idiopathic GH Deficiency. Journal of Clinical Endocrinology and Metabolism, 2015, 100, 3494-3501.	3.6	2
51	Treatment with medium chain fatty acids milk of CD36-deficient preschool children. Nutrition, 2018, 50, 45-48.	2.4	2
52	Possible critical region associated with late-onset spasms in 17p13.1â€"p13.2 microdeletion syndrome: a report of two new cases and review of the literature. Epileptic Disorders, 2022, 24, 1-5.	1.3	2
53	Blood asymmetric dimethylarginine and nitrite/nitrate concentrations in short-stature children born small for gestational age with and without growth hormone therapy. Journal of International Medical Research, 2018, 46, 761-772.	1.0	1
54	Longitudinal Glycaemic Profiles during Remission in 6q24-Related Transient Neonatal Diabetes Mellitus. Hormone Research in Paediatrics, 2021, 94, 229-234.	1.8	1

#	Article	IF	Citations
55	A 36-year-old Man with Repeated Short-term Transient Hyperammonemia and Impaired Consciousness with a Confirmed Carbamoyl Phosphate Synthase 1 Gene Monoallelic Mutation. Internal Medicine, 2022, , .	0.7	1
56	Accelerated pubertal onset in short children with delayed bone age. Journal of Pediatric Endocrinology and Metabolism, 2022, 35, 163-168.	0.9	0