Thipwimol Tim-Aroon

List of Publications by Year
in descending order

31	184	1163117 8	1199594 12
papers	citations	h-index	g-index

33	33	33	557
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sleep Disorder: An Overlooked Manifestation of Glucose Transporter Type-1 Deficiency Syndrome. Neuropediatrics, 2022, 53, 129-132.	0.6	3
2	Etiology and outcome of acute recurrent pancreatitis and chronic pancreatitis. Pediatrics International, 2022, 64, e15145.	0.5	3
3	A generation of human induced pluripotent stem cell line (MUi031-A) from a type-3 Gaucher disease patient carrying homozygous mutation on GBA1 gene. Stem Cell Research, 2022, 60, 102698.	0.7	3
4	Epidermolysis Bullosa With Congenital Absence of Skin: Congenital Corneal Cloudiness and Esophagogastric Obstruction Including Extended Genotypic Spectrum of PLEC, LAMC2, ITGB4 and COL7A1. Frontiers in Genetics, 2022, 13, 847150.	2.3	3
5	A novel TCN2 mutation with unusual clinical manifestations of hemolytic crisis and unexplained metabolic acidosis: expanding the genotype and phenotype of transcobalamin II deficiency. BMC Pediatrics, 2022, 22, 233.	1.7	2
6	Comparison of 24-Hour Recall and 3-Day Food Records during the Complementary Feeding Period in Thai Infants and Evaluation of Plasma Amino Acids as Markers of Protein Intake. Nutrients, 2021, 13, 653.	4.1	6
7	Central Precocious Puberty in a Boy with Pseudohypoparathyroidism Type 1A due to a Novel GNAS Variant, Who Had Congenital Hypothyroidism as the First Manifestation. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2021, .	0.9	0
8	Infantile onset Sandhoff disease: clinical manifestation and a novel common mutation in Thai patients. BMC Pediatrics, 2021, 21, 22.	1.7	8
9	Gaucher disease: clinical phenotypes and refining GBA mutational spectrum in Thai patients. Orphanet Journal of Rare Diseases, 2021, 16, 519.	2.7	4
10	Perspectives of adults with Klinefelter syndrome, unaffected adolescent males, and parents of affected children toward diagnosis disclosure: a Thai experience. Journal of Community Genetics, 2020, 11, 171-181.	1.2	4
11	Successful parathyroidectomy with intraâ€operative parathyroid hormone monitoring in a neonate with severe primary hyperparathyroidism caused by homozygous mutation in CASR gene. Journal of Paediatrics and Child Health, 2020, 56, 1144-1146.	0.8	1
12	Novel SOX10 Mutations in Waardenburg Syndrome: Functional Characterization and Genotype-Phenotype Analysis. Frontiers in Genetics, 2020, 11, 589784.	2.3	10
13	MITF variants cause nonsyndromic sensorineural hearing loss with autosomal recessive inheritance. Scientific Reports, 2020, 10, 12712.	3.3	9
14	Kabuki syndrome with midgut malrotation and hyperinsulinemic hypoglycemia: A rare coâ€occurrence from Thailand. American Journal of Medical Genetics, Part A, 2020, 182, 1873-1876.	1.2	1
15	Klinefelter Syndrome Mosaicism 46,XX/47,XXY: A New Case and Literature Review. Journal of Pediatric Genetics, 2020, 09, 221-226.	0.7	2
16	DUOX2 variants are a frequent cause of congenital primary hypothyroidism in Thai patients. Endocrine Connections, 2020, 9, 1121-1134.	1.9	12
17	Growth charts for Thai children with Prader-Willi syndrome aged 0â€“18â€‰years. Orphanet Journal of Rare Diseases, 2020, 15, 111.	2.7	2
18	Etiologies, Prognostic Factors, and Outcomes of Pediatric Acute Liver Failure in Thailand. Pediatric Gastroenterology, Hepatology and Nutrition, 2020, 23, 539.	1.2	3

#	Article	IF	Citations
19	Clinical delineation of 18q11â€q12 microdeletion: Intellectual disability, speech and behavioral disorders, and conotruncal heart defects. Molecular Genetics & Genomic Medicine, 2019, 7, e896.	1.2	5
20	Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand. BMC Medical Genetics, 2019, 20, 156.	2.1	7
21	Two infants with abetalipoproteinemia: Classic versus atypical presentation. Pediatrics International, 2019, 61, 508-509.	0.5	2
22	p.X654R IDUA variant among Thai individuals with intermediate mucopolysaccharidosis type I and its residual activity as demonstrated in COS-7 cells. Annals of Human Genetics, 2018, 82, 150-157.	0.8	6
23	Case 3: Newborn with Asymmetric Crying Face. NeoReviews, 2018, 19, e304-e307.	0.8	0
24	1q21.3 deletion involving GATAD2B: An emerging recurrent microdeletion syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 766-770.	1.2	10
25	Clinical improvement of renal amyloidosis in a patient with systemic-onset juvenile idiopathic arthritis who received tocilizumab treatment: a case report and literature review. BMC Nephrology, 2017, 18, 159.	1.8	8
26	Chromosomal microarray analysis in a cohort of underrepresented population identifies SERINC2 as a novel candidate gene for autism spectrum disorder. Scientific Reports, 2017, 7, 12096.	3.3	21
27	Molecular analysis of the novel IDS allele in a Thai family with mucopolysaccharidosis type II: The c.928C>T (p.Gln310*) transcript is sensitive to nonsense-mediated mRNA decay. Experimental and Therapeutic Medicine, 2017, 13, 2989-2996.	1.8	1
28	The Value of Comprehensive Thyroid Function Testing and Family History for Early Diagnosis of MCT8 Deficiency. Clinical Pediatrics, 2016, 55, 286-289.	0.8	4
29	Stopping Parenteral Nutrition for 3 Hours Reduces False Positives in Newborn Screening. Journal of Pediatrics, 2015, 167, 312-316.	1.8	10
30	Novel SMC1A frameshift mutations in children with developmental delay and epilepsy. European Journal of Medical Genetics, 2015, 58, 562-568.	1.3	26
31	A new case of maternal lupusâ€associated chondrodysplasia punctata with extensive spinal anomalies. American Journal of Medical Genetics, Part A, 2011, 155, 1487-1491.	1.2	8