

Thipwimol Tim-Aroon

List of Publications by Year in descending order

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Version: 2024-02-01

31
papers

184
citations

1163117

8
h-index

1199594

12
g-index

33
all docs

33
docs citations

33
times ranked

557
citing authors

#	ARTICLE	IF	CITATIONS
1	Novel SMC1A frameshift mutations in children with developmental delay and epilepsy. <i>European Journal of Medical Genetics</i> , 2015, 58, 562-568.	1.3	26
2	Chromosomal microarray analysis in a cohort of underrepresented population identifies SERINC2 as a novel candidate gene for autism spectrum disorder. <i>Scientific Reports</i> , 2017, 7, 12096.	3.3	21
3	DUOX2 variants are a frequent cause of congenital primary hypothyroidism in Thai patients. <i>Endocrine Connections</i> , 2020, 9, 1121-1134.	1.9	12
4	Stopping Parenteral Nutrition for 3 Hours Reduces False Positives in Newborn Screening. <i>Journal of Pediatrics</i> , 2015, 167, 312-316.	1.8	10
5	1q21.3 deletion involving <i>GATAD2B</i> : An emerging recurrent microdeletion syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 766-770.	1.2	10
6	Novel SOX10 Mutations in Waardenburg Syndrome: Functional Characterization and Genotype-Phenotype Analysis. <i>Frontiers in Genetics</i> , 2020, 11, 589784.	2.3	10
7	MITF variants cause nonsyndromic sensorineural hearing loss with autosomal recessive inheritance. <i>Scientific Reports</i> , 2020, 10, 12712.	3.3	9
8	A new case of maternal lupus-associated chondrodysplasia punctata with extensive spinal anomalies. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1487-1491.	1.2	8
9	Clinical improvement of renal amyloidosis in a patient with systemic-onset juvenile idiopathic arthritis who received tocilizumab treatment: a case report and literature review. <i>BMC Nephrology</i> , 2017, 18, 159.	1.8	8
10	Infantile onset Sandhoff disease: clinical manifestation and a novel common mutation in Thai patients. <i>BMC Pediatrics</i> , 2021, 21, 22.	1.7	8
11	Clinical course, mutations and its functional characteristics of infantile-onset Pompe disease in Thailand. <i>BMC Medical Genetics</i> , 2019, 20, 156.	2.1	7
12	p.X654R IDUA variant among Thai individuals with intermediate mucopolysaccharidosis type I and its residual activity as demonstrated in COS-7 cells. <i>Annals of Human Genetics</i> , 2018, 82, 150-157.	0.8	6
13	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. <i>Nutrients</i> , 2021, 13, 653.	4.1	6
14	Clinical delineation of 18q11-q12 microdeletion: Intellectual disability, speech and behavioral disorders, and conotruncal heart defects. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e896.	1.2	5
15	The Value of Comprehensive Thyroid Function Testing and Family History for Early Diagnosis of MCT8 Deficiency. <i>Clinical Pediatrics</i> , 2016, 55, 286-289.	0.8	4
16	Perspectives of adults with Klinefelter syndrome, unaffected adolescent males, and parents of affected children toward diagnosis disclosure: a Thai experience. <i>Journal of Community Genetics</i> , 2020, 11, 171-181.	1.2	4
17	Gaucher disease: clinical phenotypes and refining GBA mutational spectrum in Thai patients. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 519.	2.7	4
18	Sleep Disorder: An Overlooked Manifestation of Glucose Transporter Type-1 Deficiency Syndrome. <i>Neuropediatrics</i> , 2022, 53, 129-132.	0.6	3

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19	Etiologies, Prognostic Factors, and Outcomes of Pediatric Acute Liver Failure in Thailand. <i>Pediatric Gastroenterology, Hepatology and Nutrition</i> , 2020, 23, 539.	1.2	3
20	Etiology and outcome of acute recurrent pancreatitis and chronic pancreatitis. <i>Pediatrics International</i> , 2022, 64, e15145.	0.5	3
21	A generation of human induced pluripotent stem cell line (MUI031-A) from a type-3 Gaucher disease patient carrying homozygous mutation on GBA1 gene. <i>Stem Cell Research</i> , 2022, 60, 102698.	0.7	3
22	Epidermolysis Bullosa With Congenital Absence of Skin: Congenital Corneal Cloudiness and Esophagogastric Obstruction Including Extended Genotypic Spectrum of PLEC, LAMC2, ITGB4 and COL7A1. <i>Frontiers in Genetics</i> , 2022, 13, 847150.	2.3	3
23	Two infants with abetalipoproteinemia: Classic versus atypical presentation. <i>Pediatrics International</i> , 2019, 61, 508-509.	0.5	2
24	Klinefelter Syndrome Mosaicism 46,XX/47,XXY: A New Case and Literature Review. <i>Journal of Pediatric Genetics</i> , 2020, 09, 221-226.	0.7	2
25	Growth charts for Thai children with Prader-Willi syndrome aged 0-18 years. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 111.	2.7	2
26	A novel TCN2 mutation with unusual clinical manifestations of hemolytic crisis and unexplained metabolic acidosis: expanding the genotype and phenotype of transcobalamin II deficiency. <i>BMC Pediatrics</i> , 2022, 22, 233.	1.7	2
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. <i>Experimental and Therapeutic Medicine</i> , 2017, 13, 2989-2996.	1.8	1
28	Successful parathyroidectomy with intraoperative parathyroid hormone monitoring in a neonate with severe primary hyperparathyroidism caused by homozygous mutation in CASR gene. <i>Journal of Paediatrics and Child Health</i> , 2020, 56, 1144-1146.	0.8	1
29	Kabuki syndrome with midgut malrotation and hyperinsulinemic hypoglycemia: A rare occurrence from Thailand. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1873-1876.	1.2	1
30	Case 3: Newborn with Asymmetric Crying Face. <i>NeoReviews</i> , 2018, 19, e304-e307.	0.8	0
31	Central Precocious Puberty in a Boy with Pseudohypoparathyroidism Type 1A due to a Novel <i>GNAS</i> Variant, Who Had Congenital Hypothyroidism as the First Manifestation. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2021, .	0.9	0