Chupong Ittiwut

List of Publications by Year
in descending order

30	254	1162367 8	1058022 14
papers	citations	h-index	g-index

31	31	31	491
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Phenotypic heterogeneity and genotypic spectrum of inborn errors of immunity identified through whole exome sequencing in a Thai patient cohort. Pediatric Allergy and Immunology, 2022, 33, .	1.1	2
2	Novel CD55 Mutation Associated With Severe Small Bowel Ulceration Mimicking Inflammatory Bowel Disease in a Pair of Siblings. Inflammatory Bowel Diseases, 2022, 28, 1458-1461.	0.9	4
3	Novel de novo mutation substantiates ATP6V0C as a gene causing epilepsy with intellectual disability. Brain and Development, 2021, 43, 490-494.	0.6	5
4	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. Nephron, 2021, 145, 311-316.	0.9	3
5	Rapid exome sequencing as the firstâ€tier investigation for diagnosis of acutely and severely ill children and adults in Thailand. Clinical Genetics, 2021, 100, 100-105.	1.0	12
6	Severe neonatal haemolytic anaemia caused by compound heterozygous KLF1 mutations: report of four families and literature review. British Journal of Haematology, 2021, 194, 626-634.	1.2	9
7	Diagnosis of Hyper IgM syndrome in a Previously Healthy Adolescent Boy Presented with Cutaneous and Cerebral Cryptococcosis. Pediatric Infectious Disease Journal, 2021, 40, e18-e20.	1.1	3
8	Coinherited Hemoglobin H/Constant Spring Disease and Heterozygous Hemoglobin Tak Causing Severe Hemolytic Anemia in a Thai Boy. Journal of Pediatric Hematology/Oncology, 2021, 43, e723-e726.	0.3	1
9	Compound Heterozygous PGM3 Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. Journal of Clinical Immunology, 2020, 40, 227-231.	2.0	4
10	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. Neuromuscular Disorders, 2020, 30, 851-858.	0.3	7
11	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. European Journal of Medical Genetics, 2020, 63, 104086.	0.7	6
12	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	1.0	3
13	A case of GABRA5-related developmental and epileptic encephalopathy with response to a combination of antiepileptic drugs and a GABAering agent. Brain and Development, 2020, 42, 546-550.	0.6	1
14	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. Journal of Clinical Neuroscience, 2020, 72, 31-38.	0.8	8
15	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	1.0	8
16	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. Medicine (United States), 2020, 99, e23275.	0.4	7
17	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. Case Reports in Dermatology, 2020, 12, 241-248.	0.3	1
18	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushingâ€™s Syndrome. Hormone Research in Paediatrics, 2019, 92, 196-202.	0.8	3

#	Article	IF	Citations
19	Discrepancy in the degree of polycythemia in a family with a novel nonsense EPOR mutation. International Journal of Hematology, 2019, 110, 640-641.	0.7	0
20	Female-restricted syndromic intellectual disability in a patient from Thailand. , 2019, 179, 758-761.		8
21	Whole exome sequencing revealed mutations in FBXL4, UNC80, and ADK in Thai patients with severe intellectual disabilities. Gene, 2019, 696, 21-27.	1.0	12
22	Novel mutations in <scp>SPTA</scp>1 and <scp>SPTB</scp> identified by whole exome sequencing in eight Thai families with hereditary pyropoikilocytosis presenting with severe fetal and neonatal anaemia. British Journal of Haematology, 2019, 185, 578-582.	1.2	16
23	ATP1A3-related Epilepsy: Report of Six Cases and Literature-based Analysis of Treatment Response. Neuropediatrics, 2019, 50, .	0.3	0
24	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. Gene, 2018, 679, 377-381.	1.0	16
25	Novel mutations in Thai patients with glanzmann thrombasthenia. European Journal of Haematology, 2017, 99, 520-524.	1.1	3
26	Novel Mutations, Including a Large Deletion in theARSBGene, Causing Mucopolysaccharidosis Type VI. Genetic Testing and Molecular Biomarkers, 2017, 21, 58-62.	0.3	8
27	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. BMC Medical Genetics, 2017, 18, 102.	2.1	14
28	Bilateral Femoral Neck Fractures in Cerebrotendinous Xanthomatosis Treated by Hip Arthroplasties: The First Case Report and Literature Review. Journal of Orthopaedic Case Reports, 2017, 7, 54-58.	0.1	2
29	NUDT15 c.415C>T increases risk of 6-mercaptopurine induced myelosuppression during maintenance therapy in children with acute lymphoblastic leukemia. Haematologica, 2016, 101, e24-e26.	1.7	76
30	Variants of the CDH1 (E-Cadherin) Gene Associated with Oral Clefts in the Thai Population. Genetic Testing and Molecular Biomarkers, 2016, 20, 406-409.	0.3	12