Chupong Ittiwut

List of Publications by Year in Descending Order

27 papers	134 citations	6 h-index	11 g-index
31 ext. papers	204 ext. citations	2.9 avg, IF	2.38 L-index

#	Paper	IF	Citations
27	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	3.4	0
26	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	1.2	
25	Rapid exome sequencing as the first-tier investigation for diagnosis of acutely and severely ill children and adults in Thailand. *Clinical Genetics*, **2021**, 100, 100-105	4	4
24	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	4.5	1
23	Novel de novo mutation substantiates ATP6V0C as a gene causing epilepsy with intellectual disability. *Brain and Development*, **2021**, 43, 490-494	2.2	1
22	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. *Nephron*, **2021**, 145, 311-316	3.3	0
21	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	2.2	0
20	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. *Journal of Clinical Neuroscience*, **2020**, 72, 31-38	2.2	1
19	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. *Gene*, **2020**, 749, 144709	3.8	4
18	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. *Medicine (United States)*, **2020**, 99, e23275	1.8	1
17	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. *Case Reports in Dermatology*, **2020**, 12, 241-248	1.1	
16	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	5.7	2
15	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. *Neuromuscular Disorders*, **2020**, 30, 851-858	2.9	1
14	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. *European Journal of Medical Genetics*, **2020**, 63, 104086	2.6	4
13	Clinical and molecular characteristics of Thai patients with related neutropaenia. *Journal of Clinical Pathology*, **2020**,	3.9	1
12	Female-restricted syndromic intellectual disability in a patient from Thailand. *American Journal of Medical Genetics, Part A*, **2019**, 179, 758-761	2.5	2
11	Whole exome sequencing revealed mutations in FBXL4, UNC80, and ADK in Thai patients with severe intellectual disabilities. *Gene*, **2019**, 696, 21-27	3.8	7

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10	A Novel GNAS Mutation Causing Isolated Infantile Cushing's Syndrome. *Hormone Research in Paediatrics*, **2019**, 92, 196-202	3.3	0
9	Discrepancy in the degree of polycythemia in a family with a novel nonsense EPOR mutation. *International Journal of Hematology*, **2019**, 110, 640-641	2.3	
8	Novel mutations in SPTA1 and SPTB 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	4.5	8
7	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. *Gene*, **2018**, 679, 377-381	3.8	7
6	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. *BMC Medical Genetics*, **2017**, 18, 102	2.1	7
5	Novel mutations in Thai patients with glanzmann thrombasthenia. *European Journal of Haematology*, **2017**, 99, 520-524	3.8	3
4	Novel Mutations, Including a Large Deletion in the ARSB Gene, Causing Mucopolysaccharidosis Type VI. *Genetic Testing and Molecular Biomarkers*, **2017**, 21, 58-62	1.6	6
3	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	0.3	2
2	Variants of the CDH1 (E-Cadherin) Gene Associated with Oral Clefts in the Thai Population. *Genetic Testing and Molecular Biomarkers*, **2016**, 20, 406-9	1.6	12
1	NUDT15 c.415C>T increases risk of 6-mercaptopurine induced myelosuppression during maintenance therapy in children with acute lymphoblastic leukemia. *Haematologica*, **2016**, 101, e24-6	6.6	60