## **Chupong Ittiwut**

## List of Publications by Citations

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

#	Paper	IF	Citations
27	NUDT15 c.415C>T increases risk of 6-mercaptopurine induced myelosuppression during maintenance therapy in children with acute lymphoblastic leukemia. <i>Haematologica</i> , <b>2016</b> , 101, e24-6	6.6	60
26	Variants of the CDH1 (E-Cadherin) Gene Associated with Oral Clefts in the Thai Population. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2016</b> , 20, 406-9	1.6	12
25	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. <i>British Journal of Haematology</i> , <b>2019</b> , 185, 578-582	4.5	8
24	Whole exome sequencing revealed mutations in FBXL4, UNC80, and ADK in Thai patients with severe intellectual disabilities. <i>Gene</i> , <b>2019</b> , 696, 21-27	3.8	7
23	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. <i>BMC Medical Genetics</i> , <b>2017</b> , 18, 102	2.1	7
22	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. <i>Gene</i> , <b>2018</b> , 679, 377-381	3.8	7
21	Novel Mutations, Including a Large Deletion in the ARSB Gene, Causing Mucopolysaccharidosis Type VI. <i>Genetic Testing and Molecular Biomarkers</i> , <b>2017</b> , 21, 58-62	1.6	6
20	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. <i>Gene</i> , <b>2020</b> , 749, 144709	3.8	4
19	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. <i>European Journal of Medical Genetics</i> , <b>2020</b> , 63, 104086	2.6	4
18	Rapid exome sequencing as the first-tier investigation for diagnosis of acutely and severely ill children and adults in Thailand. <i>Clinical Genetics</i> , <b>2021</b> , 100, 100-105	4	4
17	Novel mutations in Thai patients with glanzmann thrombasthenia. <i>European Journal of Haematology</i> , <b>2017</b> , 99, 520-524	3.8	3
16	Female-restricted syndromic intellectual disability in a patient from Thailand. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 758-761	2.5	2
15	Bilateral Femoral Neck Fractures in Cerebrotendinous Xanthomatosis Treated by Hip Arthroplasties: The First Case Report and Literature Review. <i>Journal of Orthopaedic Case Reports</i> , <b>2017</b> , 7, 54-58	0.3	2
14	Compound Heterozygous PGM3 Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. <i>Journal of Clinical Immunology</i> , <b>2020</b> , 40, 227-231	5.7	2
13	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. <i>Journal of Clinical Neuroscience</i> , <b>2020</b> , 72, 31-38	2.2	1
12	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. <i>Medicine (United States)</i> , <b>2020</b> , 99, e23275	1.8	1
11	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. <i>Neuromuscular Disorders</i> , <b>2020</b> , 30, 851-858	2.9	1

## LIST OF PUBLICATIONS

10	Clinical and molecular characteristics of Thai patients with related neutropaenia. <i>Journal of Clinical Pathology</i> , <b>2020</b> ,	3.9	1
9	Severe neonatal haemolytic anaemia caused by compound heterozygous KLF1 mutations: report of four families and literature review. <i>British Journal of Haematology</i> , <b>2021</b> , 194, 626-634	4.5	1
8	Novel de novo mutation substantiates ATP6V0C as a gene causing epilepsy with intellectual disability. <i>Brain and Development</i> , <b>2021</b> , 43, 490-494	2.2	1
7	A case of GABRA5-related developmental and epileptic encephalopathy with response to a combination of antiepileptic drugs and a GABAering agent. <i>Brain and Development</i> , <b>2020</b> , 42, 546-550	2.2	0
6	A Novel GNAS Mutation Causing Isolated Infantile Cushing Syndrome. <i>Hormone Research in Paediatrics</i> , <b>2019</b> , 92, 196-202	3.3	O
5	Diagnosis of Hyper IgM syndrome in a Previously Healthy Adolescent Boy Presented with Cutaneous and Cerebral Cryptococcosis. <i>Pediatric Infectious Disease Journal</i> , <b>2021</b> , 40, e18-e20	3.4	O
4	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. <i>Nephron</i> , <b>2021</b> , 145, 311-316	3.3	O
3	Discrepancy in the degree of polycythemia in a family with a novel nonsense EPOR mutation. <i>International Journal of Hematology</i> , <b>2019</b> , 110, 640-641	2.3	
2	Coinherited Hemoglobin H/Constant Spring Disease and Heterozygous Hemoglobin Tak Causing Severe Hemolytic Anemia in a Thai Boy. <i>Journal of Pediatric Hematology/Oncology</i> , <b>2021</b> , 43, e723-e726	1.2	
1	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. <i>Case Reports in Dermatology</i> , <b>2020</b> , 12, 241-248	1.1	