Rungnapa Ittiwut

List of Publications by Year
in descending order

17 papers	155 citations	1464605 7 h-index	1427216 11 g-index
17 all docs	17 docs citations	17 times ranked	290 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	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
5	Trinucleotide repeat expansion in the transcription factor 4 (TCF4) gene in Thai patients with Fuchs endothelial corneal dystrophy. Eye, 2020, 34, 880-885.	1.1	7
6	A novel deletion in the fibrinogen beta chain (FGB) gene causing hypofibrinogenemia. Thrombosis Research, 2020, 186, 26-29.	0.8	1
7	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	1.0	3
8	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. Medicine (United States), 2020, 99, e23275.	0.4	7
9	Identification and Functional Analysis of Six DAX1 Mutations in Patients With X-Linked Adrenal Hypoplasia Congenita. Journal of the Endocrine Society, 2019, 3, 171-180.	0.1	13
10	Mutations in Kinesin family member 6 reveal specific role in ependymal cell ciliogenesis and human neurological development. PLoS Genetics, 2018, 14, e1007817.	1.5	45
11	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. Gene, 2018, 679, 377-381.	1.0	16
12	rs11567842 SNP in SLC13A2 gene associates with hypocitraturia in Thai patients with nephrolithiasis. Genes and Genomics, 2018, 40, 965-972.	0.5	4
13	Epidemiology of cleft lip with or without cleft palate in Thais. Asian Biomedicine, 2017, 10, 335-338.	0.2	6
14	Novel mutations in Thai patients with glanzmann thrombasthenia. European Journal of Haematology, 2017, 99, 520-524.	1.1	3
15	Novel Mutations, Including a Large Deletion in theARSBGene, Causing Mucopolysaccharidosis Type VI. Genetic Testing and Molecular Biomarkers, 2017, 21, 58-62.	0.3	8
16	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
17	FOXE1 mutations in Thai patients with oral clefts. Genetical Research, 2013, 95, 133-137.	0.3	7