

# Rungnapa Ittiwut

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/6003978/publications.pdf>

Version: 2024-02-01

17  
papers

155  
citations

1306789

7  
h-index

1281420

11  
g-index

17  
all docs

17  
docs citations

17  
times ranked

272  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in Kinesin family member 6 reveal specific role in ependymal cell ciliogenesis and human neurological development. <i>PLoS Genetics</i> , 2018, 14, e1007817.	1.5	45
2	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. <i>Gene</i> , 2018, 679, 377-381.	1.0	16
3	Identification and Functional Analysis of Six DAX1 Mutations in Patients With X-Linked Adrenal Hypoplasia Congenita. <i>Journal of the Endocrine Society</i> , 2019, 3, 171-180.	0.1	13
4	Variants of the <i>CDH1</i> (E-Cadherin) Gene Associated with Oral Clefts in the Thai Population. <i>Genetic Testing and Molecular Biomarkers</i> , 2016, 20, 406-409.	0.3	12
5	Rapid exome sequencing as the first-tier investigation for diagnosis of acutely and severely ill children and adults in Thailand. <i>Clinical Genetics</i> , 2021, 100, 100-105.	1.0	12
6	Novel Mutations, Including a Large Deletion in the <i>ARSB</i> Gene, Causing Mucopolysaccharidosis Type VI. <i>Genetic Testing and Molecular Biomarkers</i> , 2017, 21, 58-62.	0.3	8
7	FOXE1 mutations in Thai patients with oral clefts. <i>Genetical Research</i> , 2013, 95, 133-137.	0.3	7
8	Trinucleotide repeat expansion in the transcription factor 4 (TCF4) gene in Thai patients with Fuchs endothelial corneal dystrophy. <i>Eye</i> , 2020, 34, 880-885.	1.1	7
9	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. <i>Medicine (United States)</i> , 2020, 99, e23275.	0.4	7
10	Epidemiology of cleft lip with or without cleft palate in Thais. <i>Asian Biomedicine</i> , 2017, 10, 335-338.	0.2	6
11	Novel de novo mutation substantiates ATP6VOC as a gene causing epilepsy with intellectual disability. <i>Brain and Development</i> , 2021, 43, 490-494.	0.6	5
12	rs11567842 SNP in <i>SLC13A2</i> gene associates with hypocitraturia in Thai patients with nephrolithiasis. <i>Genes and Genomics</i> , 2018, 40, 965-972.	0.5	4
13	Novel <i>CD55</i> Mutation Associated With Severe Small Bowel Ulceration Mimicking Inflammatory Bowel Disease in a Pair of Siblings. <i>Inflammatory Bowel Diseases</i> , 2022, 28, 1458-1461.	0.9	4
14	Novel mutations in Thai patients with glanzmann thrombasthenia. <i>European Journal of Haematology</i> , 2017, 99, 520-524.	1.1	3
15	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. <i>Journal of Clinical Pathology</i> , 2020, , jclinpath-2020-207139.	1.0	3
16	Phenotypic heterogeneity and genotypic spectrum of inborn errors of immunity identified through whole exome sequencing in a Thai patient cohort. <i>Pediatric Allergy and Immunology</i> , 2022, 33, .	1.1	2
17	A novel deletion in the fibrinogen beta chain (FGB) gene causing hypofibrinogenemia. <i>Thrombosis Research</i> , 2020, 186, 26-29.	0.8	1