## Kanya Suphapeetiporn

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	Genotype–phenotype correlation and expansion of orodental anomalies in LTBP3-related disorders. Molecular Genetics and Genomics, 2019, 294, 773-787.	1.0	24
2	The Thai reference exome (Tâ€REx) variant database. Clinical Genetics, 2021, 100, 703-712.	1.0	24
3	A novel <i><scp>GJA</scp>1</i> mutation in oculodentodigital dysplasia with extensive loss of enamel. Oral Diseases, 2017, 23, 795-800.	1.5	23
4	Dental properties, ultrastructure, and pulp cells associated with a novel <i><scp>DSPP</scp></i> mutation. Oral Diseases, 2018, 24, 619-627.	1.5	21
5	A novel <i>PITX2</i> mutation in nonâ€syndromic orodental anomalies. Oral Diseases, 2018, 24, 611-618.	1.5	21
6	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. BMC Medical Genetics, 2017, 18, 102.	2.1	14
7	Age-Related Reference Intervals for Blood Amino Acids in Thai Pediatric Population Measured by Liquid Chromatography Tandem Mass Spectrometry. Journal of Nutrition and Metabolism, 2018, 2018, 1-10.	0.7	14
8	Monoallelic <i>FGFR3</i> and Biallelic <i>ALPL</i> mutations in a Thai girl with hypochondroplasia and hypophosphatasia. American Journal of Medical Genetics, Part A, 2017, 173, 2747-2752.	0.7	13
9	The most 5′ truncating homozygous mutation of WNT1 in siblings with osteogenesis imperfecta with a variable degree of brain anomalies: a case report. BMC Medical Genetics, 2018, 19, 117.	2.1	9
10	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	1.0	8
11	Novel <i>CD55</i> 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
12	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	1.0	3
13	Generation of two human iPSC lines (MDCUi001-A and MDCUi001-B) from dermal fibroblasts of a Thai patient with X-linked osteogenesis imperfecta using integration-free Sendai virus. Stem Cell Research, 2019, 39, 101493.	0.3	2
14	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
15	Novel <scp><i>BMP1</i></scp> , <scp><i>CRTAP</i></scp> , and <scp><i>SERPINF1</i></scp> variants causing autosomal recessive osteogenesis imperfecta. Clinical Genetics, 2022, 102, 242-243.	1.0	1