Kanya Suphapeetiporn

List of Publications by Year
in descending order

15 papers	183 citations	1162367 8 h-index	1125271 13 g-index
15 all docs	15 docs citations	15 times ranked	271 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 <scp>BMP1</scp>, <scp>CRTAP</scp>, and <scp>SERPINF1</scp> variants causing autosomal recessive osteogenesis imperfecta. Clinical Genetics, 2022, 102, 242-243.	1.0	1
4	The Thai reference exome (Tâ€REx) variant database. Clinical Genetics, 2021, 100, 703-712.	1.0	24
5	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	1.0	3
6	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	1.0	8
7	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
8	Genotypeâ€“phenotype correlation and expansion of orodental anomalies in LTBP3-related disorders. Molecular Genetics and Genomics, 2019, 294, 773-787.	1.0	24
9	Dental properties, ultrastructure, and pulp cells associated with a novel <scp>DSPP</scp> mutation. Oral Diseases, 2018, 24, 619-627.	1.5	21
10	A novel PITX2 mutation in nonâ€syndromic orodental anomalies. Oral Diseases, 2018, 24, 611-618.	1.5	21
11	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
12	The most 5â€² 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
13	A novel <scp>GJA</scp>1 mutation in oculodentodigital dysplasia with extensive loss of enamel. Oral Diseases, 2017, 23, 795-800.	1.5	23
14	Monoallelic FGFR3 and Biallelic ALPL mutations in a Thai girl with hypochondroplasia and hypophosphatasia. American Journal of Medical Genetics, Part A, 2017, 173, 2747-2752.	0.7	13
15	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. BMC Medical Genetics, 2017, 18, 102.	2.1	14