

Kanya Suphapeetiporn

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

183
citations

1163065

8
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1125717

13
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15
all docs

15
docs citations

15
times ranked

271
citing authors

#	ARTICLE	IF	CITATIONS
1	Genotype-phenotype correlation and expansion of orodontal anomalies in LTBP3-related disorders. <i>Molecular Genetics and Genomics</i> , 2019, 294, 773-787.	2.1	24
2	The Thai reference exome (Tâ€REx) variant database. <i>Clinical Genetics</i> , 2021, 100, 703-712.	2.0	24
3	A novel <i><scp>GJA</scp>1</i> mutation in oculodentodigital dysplasia with extensive loss of enamel. <i>Oral Diseases</i> , 2017, 23, 795-800.	3.0	23
4	Dental properties, ultrastructure, and pulp cells associated with a novel <i><scp>DSPP</scp></i> mutation. <i>Oral Diseases</i> , 2018, 24, 619-627.	3.0	21
5	A novel <i><scp>PITX2</scp></i> mutation in nonâ€syndromic orodontal anomalies. <i>Oral Diseases</i> , 2018, 24, 611-618.	3.0	21
6	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. <i>BMC Medical Genetics</i> , 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. <i>Journal of Nutrition and Metabolism</i> , 2018, 2018, 1-10.	1.8	14
8	Monoallelic <i><scp>FGFR3</scp></i> and Biallelic <i><scp>ALPL</scp></i> mutations in a Thai girl with hypochondroplasia and hypophosphatasia. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2747-2752.	1.2	13
9	The most 5â€² truncating homozygous mutation of WNT1 in siblings with osteogenesis imperfecta with a variable degree of brain anomalies: a case report. <i>BMC Medical Genetics</i> , 2018, 19, 117.	2.1	9
10	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. <i>Gene</i> , 2020, 749, 144709.	2.2	8
11	Novel <i><scp>CD55</scp></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.	1.9	4
12	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. <i>Journal of Clinical Pathology</i> , 2020, , jclinpath-2020-207139.	2.0	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. <i>Stem Cell Research</i> , 2019, 39, 101493.	0.7	2
14	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, .	2.6	2
15	Novel <scp><i><scp>BMP1</scp></i></scp>, <scp><i><scp>CRTAP</scp></i></scp>, and <scp><i><scp>SERPINF1</scp></i></scp> variants causing autosomal recessive osteogenesis imperfecta. <i>Clinical Genetics</i> , 2022, 102, 242-243.	2.0	1