

Vorasuk Shotelersuk

List of Publications by Year in descending order

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Version: 2024-02-01

195
papers

5,626
citations

126708

33
h-index

98622

67
g-index

197
all docs

197
docs citations

197
times ranked

7498
citing authors

#	ARTICLE	IF	CITATIONS
1	PTEN regulates proliferation and osteogenesis of dental pulp cells and adipogenesis of human adipose-derived stem cells. <i>Oral Diseases</i> , 2023, 29, 735-746.	1.5	3
2	Age-related dental phenotypes and tooth characteristics of <i>FAM83H</i> -associated hypocalcified amelogenesis imperfecta. <i>Oral Diseases</i> , 2022, 28, 734-744.	1.5	3
3	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
4	Host genetic factors of COVID-19 susceptibility and disease severity in a Thai population. <i>Journal of Human Genetics</i> , 2022, 67, 295-301.	1.1	15
5	Genotypic and phenotypic landscapes of 51 pharmacogenes derived from whole-genome sequencing in a Thai population. <i>PLoS ONE</i> , 2022, 17, e0263621.	1.1	4
6	Patterns of molar agenesis associated with p.P20L and p.R77Q variants in <i>PAX9</i> . <i>European Journal of Oral Sciences</i> , 2022, 130, e12855.	0.7	3
7	Multifaceted roles of YEATS domain-containing proteins and novel links to neurological diseases. <i>Cellular and Molecular Life Sciences</i> , 2022, 79, 183.	2.4	3
8	Long-read Amplicon Sequencing of the <i>CYP21A2</i> in 48 Thai Patients With Steroid 21-Hydroxylase Deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2022, 107, 1939-1947.	1.8	2
9	Comparative transcriptome profiles of human dental pulp stem cells from maxillary and mandibular teeth. <i>Scientific Reports</i> , 2022, 12, .	1.6	0
10	Novel <i>BMP1</i> , <i>CRTAP</i> , and <i>SERPINF1</i> variants causing autosomal recessive osteogenesis imperfecta. <i>Clinical Genetics</i> , 2022, 102, 242-243.	1.0	1
11	Founder effect of the TTTCA repeat insertions in <i>SAMD12</i> causing BAFME1. <i>European Journal of Human Genetics</i> , 2021, 29, 343-348.	1.4	14
12	Tooth ultrastructure of a novel <i>COL1A2</i> mutation expanding its genotypic and phenotypic spectra. <i>Oral Diseases</i> , 2021, 27, 1257-1267.	1.5	7
13	Novel de novo mutation substantiates <i>ATP6V0C</i> as a gene causing epilepsy with intellectual disability. <i>Brain and Development</i> , 2021, 43, 490-494.	0.6	5
14	Phenotypic features of dentinogenesis imperfecta associated with osteogenesis imperfecta and <i>COL1A2</i> mutations. <i>Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology</i> , 2021, 131, 694-701.	0.2	4
15	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. <i>Nephron</i> , 2021, 145, 311-316.	0.9	3
16	Prenatal Sonographic Features of CHARGE Syndrome. <i>Diagnostics</i> , 2021, 11, 415.	1.3	5
17	Compound Heterozygosity for a Novel Frameshift Variant Causing Fatal Infantile Liver Failure and Genotype-Phenotype Correlation of <i>POLG</i> c.3286G>T Variant. <i>International Journal of Neonatal Screening</i> , 2021, 7, 9.	1.2	1
18	<i>MBTPS2</i> , a membrane bound protease, underlying several distinct skin and bone disorders. <i>Journal of Translational Medicine</i> , 2021, 19, 114.	1.8	11

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19	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
20	Expanding the genotypic spectrum of <i>PYCR2</i> and a common ancestry in Thai patients with hypomyelinating leukodystrophy 10. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3068-3073.	0.7	3
21	Severe neonatal haemolytic anaemia caused by compound heterozygous <i>KLF1</i> mutations: report of four families and literature review. <i>British Journal of Haematology</i> , 2021, 194, 626-634.	1.2	9
22	The Thai reference exome (Tâ€REx) variant database. <i>Clinical Genetics</i> , 2021, 100, 703-712.	1.0	24
23	Reduced ELANE and SLPI expression compromises dental pulp cell activity. <i>Cell Proliferation</i> , 2021, 54, e13132.	2.4	6
24	Actionable secondary findings in the 73 ACMG-recommended genes in 1559 Thai exomes. <i>Journal of Human Genetics</i> , 2021, , .	1.1	5
25	Diagnosis of Hyper IgM syndrome in a Previously Healthy Adolescent Boy Presented with Cutaneous and Cerebral Cryptococcosis. <i>Pediatric Infectious Disease Journal</i> , 2021, 40, e18-e20.	1.1	3
26	Coinherited Hemoglobin H/Constant Spring Disease and Heterozygous Hemoglobin Tak Causing Severe Hemolytic Anemia in a Thai Boy. <i>Journal of Pediatric Hematology/Oncology</i> , 2021, 43, e723-e726.	0.3	1
27	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in <i>SERPINB7</i> . <i>Case Reports in Dermatology</i> , 2021, 12, 241-248.	0.3	3
28	Trinucleotide repeat expansion in the transcription factor 4 (<i>TCF4</i>) gene in Thai patients with Fuchs endothelial corneal dystrophy. <i>Eye</i> , 2020, 34, 880-885.	1.1	7
29	Compound Heterozygous <i>PGM3</i> Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. <i>Journal of Clinical Immunology</i> , 2020, 40, 227-231.	2.0	4
30	A patient with combined pituitary hormone deficiency and osteogenesis imperfecta associated with mutations in <i>LHX4</i> and <i>COL1A2</i> . <i>Journal of Advanced Research</i> , 2020, 21, 121-127.	4.4	7
31	Turner syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 303-313.	0.7	15
32	A novel deletion in the fibrinogen beta chain (<i>FGB</i>) gene causing hypofibrinogenemia. <i>Thrombosis Research</i> , 2020, 186, 26-29.	0.8	1
33	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. <i>Neuromuscular Disorders</i> , 2020, 30, 851-858.	0.3	7
34	Phenotypic and Genotypic Features of Thai Patients With Nonsyndromic Tooth Agenesis and <i>WNT10A</i> Variants. <i>Frontiers in Physiology</i> , 2020, 11, 573214.	1.3	9
35	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. <i>European Journal of Medical Genetics</i> , 2020, 63, 104086.	0.7	6
36	A Pathogenic Variant in <i>ALPK3</i> Is Associated With an Autosomal Dominant Adult-onset Hypertrophic Cardiomyopathy. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e003127.	1.6	10

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37	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. <i>Journal of Clinical Pathology</i> , 2020, , jclinpath-2020-207139.	1.0	3
38	Double heterozygous variants in FBN1 and FBN2 in a Thai woman with Marfan and Beals syndromes. <i>European Journal of Medical Genetics</i> , 2020, 63, 103982.	0.7	1
39	A case of GABRA5-related developmental and epileptic encephalopathy with response to a combination of antiepileptic drugs and a GABAergic agent. <i>Brain and Development</i> , 2020, 42, 546-550.	0.6	1
40	Four novel mutations of FAM20A in amelogenesis imperfecta type IG and review of literature for its genotype and phenotype spectra. <i>Molecular Genetics and Genomics</i> , 2020, 295, 923-931.	1.0	16
41	A family with homozygous and heterozygous p.Gly337Ser mutations in COL1A2. <i>European Journal of Medical Genetics</i> , 2020, 63, 103896.	0.7	8
42	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. <i>Journal of Clinical Neuroscience</i> , 2020, 72, 31-38.	0.8	8
43	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. <i>Gene</i> , 2020, 749, 144709.	1.0	8
44	Severe craniofrontonasal syndrome in a male patient mosaic for a novel nonsense mutation in EFNB1. <i>European Journal of Medical Genetics</i> , 2020, 63, 103924.	0.7	5
45	Dosage Optimization of Efavirenz Based on a Population Pharmacokinetic-Pharmacogenetic Model of HIV-infected Patients in Thailand. <i>Clinical Therapeutics</i> , 2020, 42, 1234-1245.	1.1	4
46	Generation and characterization of HLA-universal platelets derived from induced pluripotent stem cells. <i>Scientific Reports</i> , 2020, 10, 8472.	1.6	35
47	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. <i>Medicine (United States)</i> , 2020, 99, e23275.	0.4	7
48	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. <i>Case Reports in Dermatology</i> , 2020, 12, 241-248.	0.3	1
49	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushing's Syndrome. <i>Hormone Research in Paediatrics</i> , 2019, 92, 196-202.	0.8	3
50	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.	0.3	2
51	Discrepancy in the degree of polycythemia in a family with a novel nonsense EPOR mutation. <i>International Journal of Hematology</i> , 2019, 110, 640-641.	0.7	0
52	TTTCA repeat insertions in an intron of YEATS2 in benign adult familial myoclonic epilepsy type 4. <i>Brain</i> , 2019, 142, 3360-3366.	3.7	83
53	Decreased osteogenic activity and mineralization of alveolar bone cells from a patient with amelogenesis imperfecta and FAM83H 1261G>T mutation. <i>Genes and Diseases</i> , 2019, 6, 391-397.	1.5	7
54	A novel mutation in COL1A2 leads to osteogenesis imperfecta/Ehlers-Danlos overlap syndrome with brachydactyly. <i>Genes and Diseases</i> , 2019, 6, 138-146.	1.5	21

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55	Genotype-phenotype correlation and expansion of orodental anomalies in LTBP3-related disorders. <i>Molecular Genetics and Genomics</i> , 2019, 294, 773-787.	1.0	24
56	Precision medicine in Thailand. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 245-253.	0.7	14
57	Female-restricted syndromic intellectual disability in a patient from Thailand. , 2019, 179, 758-761.		8
58	Whole exome sequencing revealed mutations in FBXL4, UNC80, and ADK in Thai patients with severe intellectual disabilities. <i>Gene</i> , 2019, 696, 21-27.	1.0	12
59	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 150-158.	0.7	40
60	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
61	Novel mutations in <i>SPTA1</i> and <i>SPTB</i> identified by whole exome sequencing in eight Thai families with hereditary pyropoikilocytosis presenting with severe fetal and neonatal anaemia. <i>British Journal of Haematology</i> , 2019, 185, 578-582.	1.2	16
62	Compromised alveolar bone cells in a patient with dentinogenesis imperfecta caused by DSPP mutation. <i>Clinical Oral Investigations</i> , 2019, 23, 303-313.	1.4	19
63	Carnitine palmitoyl transferase 1A deficiency in an adult with recurrent severe steato hepatitis aggravated by high pathologic or physiologic demands: A roller-coaster for internists. <i>Clinical and Molecular Hepatology</i> , 2019, 25, 412-416.	4.5	6
64	A somatic PIK3CA p.H1047L mutation in a Thai patient with isolated macrodactyly: a case report. <i>Asian Biomedicine</i> , 2019, 13, 33-36.	0.2	0
65	ATP1A3-related Epilepsy: Report of Six Cases and Literature-based Analysis of Treatment Response. <i>Neuropediatrics</i> , 2019, 50, .	0.3	0
66	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	0.7	55
67	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
68	Human asparagine synthetase associates with the mitotic spindle. <i>Biology Open</i> , 2018, 7, .	0.6	9
69	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. <i>Gene</i> , 2018, 679, 377-381.	1.0	16
70	rs11567842 SNP in SLC13A2 gene associates with hypocitraturia in Thai patients with nephrolithiasis. <i>Genes and Genomics</i> , 2018, 40, 965-972.	0.5	4
71	Amelogenesis imperfecta: A novel <i>FAM83H</i> mutation and characteristics of periodontal ligament cells. <i>Oral Diseases</i> , 2018, 24, 1522-1531.	1.5	13
72	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.	0.7	14

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73	Coleâ€Carpenter syndrome in a patient from Thailand. American Journal of Medical Genetics, Part A, 2018, 176, 1706-1710.	0.7	11
74	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
75	Widespread and debilitating hemangiomas in a patient with enchondromatosis and D-2-hydroxyglutaric aciduria. Skeletal Radiology, 2018, 47, 1577-1582.	1.2	7
76	Expanding the Oro-Dental and Mutational Spectra of Kabuki Syndrome and Expression of <i>KMT2D</i> and<i> KDM6A</i> in Human Tooth Germs. International Journal of Biological Sciences, 2018, 14, 381-389.	2.6	29
77	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
78	Epidemiology of cleft lip with or without cleft palate in Thais. Asian Biomedicine, 2017, 10, 335-338.	0.2	6
79	Mutation analysis and prenatal diagnosis of a family with Griscelli syndrome type 2: two novel mutations in the RAB27A gene. World Journal of Pediatrics, 2017, 13, 392-394.	0.8	0
80	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	0.7	103
81	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
82	Two novel compound heterozygous BMP1 mutations in a patient with osteogenesis imperfecta: a case report. BMC Medical Genetics, 2017, 18, 25.	2.1	16
83	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	0.7	75
84	Novel mutations of the SRD5A2 and AR genes in Thai patients with 46, XY disorders of sex development. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 19-26.	0.4	15
85	Novel mutations in Thai patients with glanzmann thrombasthenia. European Journal of Haematology, 2017, 99, 520-524.	1.1	3
86	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
87	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	0.7	68
88	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
89	Novel Mutations, Including a Large Deletion in the<i> ARSB</i> Gene, Causing Mucopolysaccharidosis Type VI. Genetic Testing and Molecular Biomarkers, 2017, 21, 58-62.	0.3	8
90	A novel de novo COL1A1 mutation in a Thai boy with osteogenesis imperfecta born to consanguineous parents. Genetics and Molecular Biology, 2017, 40, 763-767.	0.6	6

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91	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
92	Identification of a mitochondrial 12S rRNA A1555G mutation causing aminoglycoside-induced deafness in a large Thai family. <i>Asian Biomedicine</i> , 2017, 9, 211-215.	0.2	0
93	Bilateral Femoral Neck Fractures in Cerebrotendinous Xanthomatosis Treated by Hip Arthroplasties: The First Case Report and Literature Review. <i>Journal of Orthopaedic Case Reports</i> , 2017, 7, 54-58.	0.1	2
94	NUDT15 c.415C>T increases risk of 6-mercaptopurine induced myelosuppression during maintenance therapy in children with acute lymphoblastic leukemia. <i>Haematologica</i> , 2016, 101, e24-e26.	1.7	76
95	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. <i>Nature Communications</i> , 2016, 7, 11920.	5.8	112
96	Short stature, platyspondyly, hip dysplasia, and retinal detachment: an atypical type II collagenopathy caused by a novel mutation in the C-propeptide region of COL2A1: a case report. <i>BMC Medical Genetics</i> , 2016, 17, 96.	2.1	3
97	Splicing analysis of CYP11B1 mutation in a family affected with 11 β -hydroxylase deficiency: case report. <i>BMC Endocrine Disorders</i> , 2016, 16, 37.	0.9	9
98	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
99	Adaptive immune defects in a patient with leukocyte adhesion deficiency type III with a novel mutation in <i>FERMT3</i> . <i>Pediatric Allergy and Immunology</i> , 2016, 27, 214-217.	1.1	18
100	Genome-wide search followed by replication reveals genetic interaction of <i>CD80</i> and <i>ALOX5AP</i> associated with systemic lupus erythematosus in Asian populations. <i>Annals of the Rheumatic Diseases</i> , 2016, 75, 891-898.	0.5	28
101	A novel PCCB mutation in a Thai patient with propionic acidemia identified by exome sequencing. <i>Human Genome Variation</i> , 2015, 2, 15033.	0.4	3
102	Pharmacogenetic Testing Can Identify Patients Taking Atazanavir at Risk for Hyperbilirubinemia. <i>Journal of Acquired Immune Deficiency Syndromes (1999)</i> , 2015, 69, e36-e37.	0.9	5
103	Wiskott-Aldrich syndrome iPS cells produce megakaryocytes with defects in cytoskeletal rearrangement and proplatelet formation. <i>Thrombosis and Haemostasis</i> , 2015, 113, 792-805.	1.8	40
104	Gene-Based Meta-Analysis of Genome-Wide Association Study Data Identifies Independent Single-Nucleotide Polymorphisms in <i>ANXA6</i> as Being Associated With Systemic Lupus Erythematosus in Asian Populations. <i>Arthritis and Rheumatology</i> , 2015, 67, 2966-2977.	2.9	14
105	An Economic Evaluation of Neonatal Screening for Inborn Errors of Metabolism Using Tandem Mass Spectrometry in Thailand. <i>PLoS ONE</i> , 2015, 10, e0134782.	1.1	16
106	Meta-analysis of two Chinese populations identifies an autoimmune disease risk allele in 22q11.21 as associated with systemic lupus erythematosus. <i>Arthritis Research and Therapy</i> , 2015, 17, 67.	1.6	6
107	A Frameshift Mutation in <i>PEN-2</i> ; Causes Familial Comedones Syndrome. <i>Dermatology</i> , 2015, 231, 77-81.	0.9	10
108	HLAreporter: a tool for HLA typing from next generation sequencing data. <i>Genome Medicine</i> , 2015, 7, 25.	3.6	62

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109	Meta-analysis of GWAS on two Chinese populations followed by replication identifies novel genetic variants on the X chromosome associated with systemic lupus erythematosus. <i>Human Molecular Genetics</i> , 2015, 24, 274-284.	1.4	35
110	Novel AQP2 mutation causing congenital nephrogenic diabetes insipidus: challenges in management during infancy. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2014, 27, 193-7.	0.4	8
111	Two novel mutations including a large deletion of the <i>SLC4A11</i> gene causing autosomal recessive hereditary endothelial dystrophy. <i>British Journal of Ophthalmology</i> , 2014, 98, 1460-1462.	2.1	9
112	Absent expression of the osteoblast-specific maternally imprinted genes, <i>DLX5</i> and <i>DLX6</i> , causes split hand/split foot malformation type I. <i>Journal of Medical Genetics</i> , 2014, 51, 817-823.	1.5	21
113	Three SNPs in chromosome 11q23.3 are independently associated with systemic lupus erythematosus in Asians. <i>Human Molecular Genetics</i> , 2014, 23, 524-533.	1.4	29
114	ZRS 406A>G mutation in patients with tibial hypoplasia, polydactyly and triphalangeal first fingers. <i>Journal of Human Genetics</i> , 2014, 59, 467-470.	1.1	20
115	Genetics and genomics in Tailand: challenges and opportunities. <i>Molecular Genetics & Genomic Medicine</i> , 2014, 2, 210-216.	0.6	18
116	In vitro Correction of a Novel Splicing Alteration in the BTK Gene by Using Antisense Morpholino Oligonucleotides. <i>Archivum Immunologiae Et Therapiae Experimentalis</i> , 2014, 62, 431-436.	1.0	2
117	Germline and Somatic <i>DICER1</i> Mutations in a Pituitary Blastoma Causing Infantile-Onset Cushing's Disease. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E1487-E1492.	1.8	73
118	Expanding Phenotypic Spectrum of Familial Comedones. <i>Dermatology</i> , 2014, 228, 215-219.	0.9	9
119	Whole Genome and Exome Sequencing of Monozygotic Twins with Trisomy 21, Discordant for a Congenital Heart Defect and Epilepsy. <i>PLoS ONE</i> , 2014, 9, e100191.	1.1	42
120	Disorders with similar clinical phenotypes reveal underlying genetic interaction: <i>SATB2</i> acts as an activator of the <i>UPF3B</i> gene. <i>Human Genetics</i> , 2013, 132, 1383-1393.	1.8	24
121	Functional characterization of novel variants in the <i>CETP</i> promoter and the <i>LIPC</i> gene in subjects with hyperalphalipoproteinemia. <i>Clinica Chimica Acta</i> , 2013, 416, 92-95.	0.5	2
122	Meta-analysis Followed by Replication Identifies Loci in or near <i>CDKN1B</i> , <i>TET3</i> , <i>CD80</i> , <i>DRAM1</i> , and <i>ARID5B</i> as Associated with Systemic Lupus Erythematosus in Asians. <i>American Journal of Human Genetics</i> , 2013, 92, 41-51.	2.6	184
123	A newly identified locus for benign adult familial myoclonic epilepsy on chromosome 3q26.32-3q28. <i>European Journal of Human Genetics</i> , 2013, 21, 225-228.	1.4	55
124	<i>DcR3</i> Mutations in Patients with Juvenile-onset Systemic Lupus Erythematosus Lead to Enhanced Lymphocyte Proliferation. <i>Journal of Rheumatology</i> , 2013, 40, 1316-1326.	1.0	5
125	<i>MLL2</i> and <i>KDM6A</i> mutations in patients with Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2234-2243.	0.7	148
126	Novel <i>CTSK</i> mutation resulting in an entire exon 2 skipping in a Thai girl with pycnodysostosis. <i>Pediatrics International</i> , 2013, 55, 651-655.	0.2	7

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127	A common and two novel GBA mutations in Thai patients with Gaucher disease. <i>Journal of Human Genetics</i> , 2013, 58, 594-599.	1.1	17
128	FOXE1 mutations in Thai patients with oral clefts. <i>Genetical Research</i> , 2013, 95, 133-137.	0.3	7
129	FGFR1 and FGFR2 Mutations in Pfeiffer Syndrome. <i>Journal of Craniofacial Surgery</i> , 2013, 24, 150-152.	0.3	31
130	Clinical and molecular findings in Thai patients with isolated methylmalonic acidemia. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 424-429.	0.5	29
131	PDGFRa mutations in humans with isolated cleft palate. <i>European Journal of Human Genetics</i> , 2012, 20, 1058-1062.	1.4	55
132	Two novel CTNS mutations in cystinosis patients in Thailand. <i>Gene</i> , 2012, 499, 323-325.	1.0	14
133	Novel CYP11B2 mutation causing aldosterone synthase (P450c11AS) deficiency. <i>European Journal of Pediatrics</i> , 2012, 171, 1559-1562.	1.3	12
134	Primary hyperoxaluria type 1 and brachydactyly mental retardation syndrome caused by a novel mutation in <i>AGXT</i> and a terminal deletion of chromosome 2. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2124-2130.	0.7	12
135	Pharmacogenetic screening of carbamazepine-induced severe cutaneous allergic reactions. <i>Journal of Clinical Neuroscience</i> , 2011, 18, 1289-1294.	0.8	41
136	Two siblings with a novel nonsense mutation, p.R50X, in the vitamin D receptor gene. <i>Endocrine</i> , 2011, 40, 62-66.	1.1	8
137	PTPRF is disrupted in a patient with syndromic amastia. <i>BMC Medical Genetics</i> , 2011, 12, 46.	2.1	11
138	ELF1 is associated with systemic lupus erythematosus in Asian populations. <i>Human Molecular Genetics</i> , 2011, 20, 601-607.	1.4	78
139	A novel p.E276K IDUA mutation decreasing β -L-iduronidase activity causes mucopolysaccharidosis type I. <i>Molecular Vision</i> , 2011, 17, 456-60.	1.1	7
140	Concurrent bilateral pheochromocytoma and thoracic paraganglioma during pregnancy. <i>Endocrine</i> , 2010, 37, 261-264.	1.1	8
141	<i>HLA-B*1502</i> screening: Time to clinical practice. <i>Epilepsia</i> , 2010, 51, 936-938.	2.6	28
142	Functional Characterization of Vasopressin Receptor 2 Mutations Causing Partial and Complete Congenital Nephrogenic Diabetes Insipidus in Thai Families. <i>Hormone Research in Paediatrics</i> , 2010, 73, 349-354.	0.8	14
143	p.D645E of Acid β -Glucosidase Is the Most Common Mutation in Thai Patients with Infantile-Onset Pompe Disease. <i>Genetic Testing and Molecular Biomarkers</i> , 2010, 14, 835-837.	0.3	11
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