Vorasuk Shotelersuk

List of Publications by Year
in descending order

195 papers	5,626 citations	117619 34 h-index	95259 68 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 adiposeâ€derived stem cells. Oral Diseases, 2023, 29, 735-746.	3.0	3
2	Ageâ€related dental phenotypes and tooth characteristics of FAM83Hâ€associated hypocalcified amelogenesis imperfecta. Oral Diseases, 2022, 28, 734-744.	3.0	3
3	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, .	2.6	2
4	Host genetic factors of COVID-19 susceptibility and disease severity in a Thai population. Journal of Human Genetics, 2022, 67, 295-301.	2.3	15
5	Genotypic and phenotypic landscapes of 51 pharmacogenes derived from whole-genome sequencing in a Thai population. PLoS ONE, 2022, 17, e0263621.	2.5	4
6	Patterns of molar agenesis associated with p.P20L and p.R77Q variants in PAX9. European Journal of Oral Sciences, 2022, 130, e12855.	1.5	3
7	Multifaceted roles of YEATS domain-containing proteins and novel links to neurological diseases. Cellular and Molecular Life Sciences, 2022, 79, 183.	5.4	3
8	Long-read Amplicon Sequencing of the CYP21A2 in 48 Thai Patients With Steroid 21-Hydroxylase Deficiency. Journal of Clinical Endocrinology and Metabolism, 2022, 107, 1939-1947.	3.6	2
9	Comparative transcriptome profiles of human dental pulp stem cells from maxillary and mandibular teeth. Scientific Reports, 2022, 12, .	3.3	0
10	Novel <scp>BMP1</scp>, <scp>CRTAP</scp>, and <scp>SERPINF1</scp> variants causing autosomal recessive osteogenesis imperfecta. Clinical Genetics, 2022, 102, 242-243.	2.0	1
11	Founder effect of the TTTCA repeat insertions in SAMD12 causing BAFME1. European Journal of Human Genetics, 2021, 29, 343-348.	2.8	14
12	Tooth ultrastructure of a novel COL1A2 mutation expanding its genotypic and phenotypic spectra. Oral Diseases, 2021, 27, 1257-1267.	3.0	7
13	Novel de novo mutation substantiates ATP6V0C as a gene causing epilepsy with intellectual disability. Brain and Development, 2021, 43, 490-494.	1.1	5
14	Phenotypic features of dentinogenesis imperfecta associated with osteogenesis imperfecta and COL1A2 mutations. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2021, 131, 694-701.	0.4	4
15	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. Nephron, 2021, 145, 311-316.	1.8	3
16	Prenatal Sonographic Features of CHARGE Syndrome. Diagnostics, 2021, 11, 415.	2.6	5
17	Compound Heterozygosity for a Novel Frameshift Variant Causing Fatal Infantile Liver Failure and Genotypeâ€“Phenotype Correlation of POLG c.3286C>T Variant. International Journal of Neonatal Screening, 2021, 7, 9.	3.2	1
18	MBTPS2, a membrane bound protease, underlying several distinct skin and bone disorders. Journal of Translational Medicine, 2021, 19, 114.	4.4	11

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19	Rapid exome sequencing as the firstâ€tier investigation for diagnosis of acutely and severely ill children and adults in Thailand. Clinical Genetics, 2021, 100, 100-105.	2.0	12
20	Expanding the genotypic spectrum of <scp>PYCR2</scp> and a common ancestry in Thai patients with hypomyelinating leukodystrophy 10. American Journal of Medical Genetics, Part A, 2021, 185, 3068-3073.	1.2	3
21	Severe neonatal haemolytic anaemia caused by compound heterozygous KLF1 mutations: report of four families and literature review. British Journal of Haematology, 2021, 194, 626-634.	2.5	9
22	The Thai reference exome (Tâ€REx) variant database. Clinical Genetics, 2021, 100, 703-712.	2.0	24
23	Reduced ELANE and SLPI expression compromises dental pulp cell activity. Cell Proliferation, 2021, 54, e13132.	5.3	6
24	Actionable secondary findings in the 73 ACMG-recommended genes in 1559 Thai exomes. Journal of Human Genetics, 2021, , .	2.3	5
25	Diagnosis of Hyper IgM syndrome in a Previously Healthy Adolescent Boy Presented with Cutaneous and Cerebral Cryptococcosis. Pediatric Infectious Disease Journal, 2021, 40, e18-e20.	2.0	3
26	Coinherited Hemoglobin H/Constant Spring Disease and Heterozygous Hemoglobin Tak Causing Severe Hemolytic Anemia in a Thai Boy. Journal of Pediatric Hematology/Oncology, 2021, 43, e723-e726.	0.6	1
27	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. Case Reports in Dermatology, 2021, 12, 241-248.	0.8	3
28	Trinucleotide repeat expansion in the transcription factor 4 (TCF4) gene in Thai patients with Fuchs endothelial corneal dystrophy. Eye, 2020, 34, 880-885.	2.1	7
29	Compound Heterozygous PGM3 Mutations in a Thai Patient with a Specific Antibody Deficiency Requiring Monthly IVIG Infusions. Journal of Clinical Immunology, 2020, 40, 227-231.	3.8	4
30	A patient with combined pituitary hormone deficiency and osteogenesis imperfecta associated with mutations in LHX4 and COL1A2. Journal of Advanced Research, 2020, 21, 121-127.	9.5	7
31	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
32	A novel deletion in the fibrinogen beta chain (FGB) gene causing hypofibrinogenemia. Thrombosis Research, 2020, 186, 26-29.	1.7	1
33	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. Neuromuscular Disorders, 2020, 30, 851-858.	0.6	7
34	Phenotypic and Genotypic Features of Thai Patients With Nonsyndromic Tooth Agenesis and WNT10A Variants. Frontiers in Physiology, 2020, 11, 573214.	2.8	9
35	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. European Journal of Medical Genetics, 2020, 63, 104086.	1.3	6
36	A Pathogenic Variant inALPK3Is Associated With an Autosomal Dominant Adult-onset Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e003127.	3.6	10

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37	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	2.0	3
38	Double heterozygous variants in FBN1 and FBN2 in a Thai woman with Marfan and Beals syndromes. European Journal of Medical Genetics, 2020, 63, 103982.	1.3	1
39	A case of GABRA5-related developmental and epileptic encephalopathy with response to a combination of antiepileptic drugs and a GABAering agent. Brain and Development, 2020, 42, 546-550.	1.1	1
40	Four novel mutations of FAM20A in amelogenesis imperfecta type IG and review of literature for its genotype and phenotype spectra. Molecular Genetics and Genomics, 2020, 295, 923-931.	2.1	16
41	A family with homozygous and heterozygous p.Gly337Ser mutations in COL1A2. European Journal of Medical Genetics, 2020, 63, 103896.	1.3	8
42	ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. Journal of Clinical Neuroscience, 2020, 72, 31-38.	1.5	8
43	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	2.2	8
44	Severe craniofrontonasal syndrome in a male patient mosaic for a novel nonsense mutation in EFNB1. European Journal of Medical Genetics, 2020, 63, 103924.	1.3	5
45	Dosage Optimization of Efavirenz Based on a Population Pharmacokineticâ€“Pharmacogenetic Model of HIV-infected Patients in Thailand. Clinical Therapeutics, 2020, 42, 1234-1245.	2.5	4
46	Generation and characterization of HLA-universal platelets derived from induced pluripotent stem cells. Scientific Reports, 2020, 10, 8472.	3.3	35
47	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. Medicine (United States), 2020, 99, e23275.	1.0	7
48	Nagashima-Type Palmoplantar Keratosis with Compound Heterozygous Mutations in SERPINB7. Case Reports in Dermatology, 2020, 12, 241-248.	0.8	1
49	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushingâ€™s Syndrome. Hormone Research in Paediatrics, 2019, 92, 196-202.	1.8	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. Stem Cell Research, 2019, 39, 101493.	0.7	2
51	Discrepancy in the degree of polycythemia in a family with a novel nonsense EPOR mutation. International Journal of Hematology, 2019, 110, 640-641.	1.6	0
52	TTTCA repeat insertions in an intron of YEATS2 in benign adult familial myoclonic epilepsy type 4. Brain, 2019, 142, 3360-3366.	7.6	83
53	Decreased osteogenic activity and mineralization of alveolar bone cells from a patient with amelogenesis imperfecta and FAM83H 1261G>T mutation. Genes and Diseases, 2019, 6, 391-397.	3.4	7
54	A novel mutation in COL1A2 leads to osteogenesis imperfecta/Ehlers-Danlos overlap syndrome with brachydactyly. Genes and Diseases, 2019, 6, 138-146.	3.4	21

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55	Genotypeâ€“phenotype correlation and expansion of orodental anomalies in LTBP3-related disorders. Molecular Genetics and Genomics, 2019, 294, 773-787.	2.1	24
56	Precision medicine in Thailand. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 245-253.	1.6	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. Gene, 2019, 696, 21-27.	2.2	12
59	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
60	Identification and Functional Analysis of Six DAX1 Mutations in Patients With X-Linked Adrenal Hypoplasia Congenita. Journal of the Endocrine Society, 2019, 3, 171-180.	0.2	13
61	Novel mutations in <scp>SPTA</scp>1 and <scp>SPTB</scp> identified by whole exome sequencing in eight Thai families with hereditary pyropoikilocytosis presenting with severe fetal and neonatal anaemia. British Journal of Haematology, 2019, 185, 578-582.	2.5	16
62	Compromised alveolar bone cells in a patient with dentinogenesis imperfecta caused by DSPP mutation. Clinical Oral Investigations, 2019, 23, 303-313.	3.0	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. Clinical and Molecular Hepatology, 2019, 25, 412-416.	8.9	6
64	A somatic PIK3CA p.H1047L mutation in a Thai patient with isolated macrodactyly: a case report. Asian Biomedicine, 2019, 13, 33-36.	0.3	0
65	ATP1A3-related Epilepsy: Report of Six Cases and Literature-based Analysis of Treatment Response. Neuropediatrics, 2019, 50, .	0.6	0
66	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
67	Mutations in Kinesin family member 6 reveal specific role in ependymal cell ciliogenesis and human neurological development. PLoS Genetics, 2018, 14, e1007817.	3.5	45
68	Human asparagine synthetase associates with the mitotic spindle. Biology Open, 2018, 7, .	1.2	9
69	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. Gene, 2018, 679, 377-381.	2.2	16
70	rs11567842 SNP in SLC13A2 gene associates with hypocitraturia in Thai patients with nephrolithiasis. Genes and Genomics, 2018, 40, 965-972.	1.4	4
71	Amelogenesis imperfecta: A novel FAM83H mutation and characteristics of periodontal ligament cells. Oral Diseases, 2018, 24, 1522-1531.	3.0	13
72	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.	1.8	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.	1.2	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.	2.0	7
76	Expanding the Oro-Dental and Mutational Spectra of Kabuki Syndrome and Expression of KMT2D and KDM6A in Human Tooth Germs. International Journal of Biological Sciences, 2018, 14, 381-389.	6.4	29
77	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
78	Epidemiology of cleft lip with or without cleft palate in Thais. Asian Biomedicine, 2017, 10, 335-338.	0.3	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.	1.8	0
80	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
81	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	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.	1.2	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.	0.9	15
85	Novel mutations in Thai patients with glanzmann thrombasthenia. European Journal of Haematology, 2017, 99, 520-524.	2.2	3
86	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
87	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
88	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.	1.2	13
89	Novel Mutations, Including a Large Deletion in theARSBGene, Causing Mucopolysaccharidosis Type VI. Genetic Testing and Molecular Biomarkers, 2017, 21, 58-62.	0.7	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.	1.3	6

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91	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. BMC Medical Genetics, 2017, 18, 102.	2.1	14
92	Identification of a mitochondrial 12S rRNA A1555G mutation causing aminoglycoside-induced deafness in a large Thai family. Asian Biomedicine, 2017, 9, 211-215.	0.3	0
93	Bilateral Femoral Neck Fractures in Cerebrotendinous Xanthomatosis Treated by Hip Arthroplasties: The First Case Report and Literature Review. Journal of Orthopaedic Case Reports, 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. Haematologica, 2016, 101, e24-e26.	3.5	76
95	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920.	12.8	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. BMC Medical Genetics, 2016, 17, 96.	2.1	3
97	Splicing analysis of CYP11B1 mutation in a family affected with 11Î²-hydroxylase deficiency: case report. BMC Endocrine Disorders, 2016, 16, 37.	2.2	9
98	Variants of the CDH1 (E-Cadherin) Gene Associated with Oral Clefts in the Thai Population. Genetic Testing and Molecular Biomarkers, 2016, 20, 406-409.	0.7	12
99	Adaptive immune defects in a patient with leukocyte adhesion deficiency type <scp>III</scp> with a novel mutation in <scp>FERMT</scp>3. Pediatric Allergy and Immunology, 2016, 27, 214-217.	2.6	18
100	Genome-wide search followed by replication reveals genetic interaction ofCD80andALOX5APassociated with systemic lupus erythematosus in Asian populations. Annals of the Rheumatic Diseases, 2016, 75, 891-898.	0.9	28
101	A novel PCCB mutation in a Thai patient with propionic acidemia identified by exome sequencing. Human Genome Variation, 2015, 2, 15033.	0.7	3
102	Pharmacogenetic Testing Can Identify Patients Taking Atazanavir at Risk for Hyperbilirubinemia. Journal of Acquired Immune Deficiency Syndromes (1999), 2015, 69, e36-e37.	2.1	5
103	Wiskott-Aldrich syndrome iPS cells produce megakaryocytes with defects in cytoskeletal rearrangement and proplatelet formation. Thrombosis and Haemostasis, 2015, 113, 792-805.	3.4	40
104	Geneâ€Based Metaâ€Analysis of Genomeâ€Wide Association Study Data Identifies Independent Singleâ€Nucleotide Polymorphisms in ANXA6 as Being Associated With Systemic Lupus Erythematosus in Asian Populations. Arthritis and Rheumatology, 2015, 67, 2966-2977.	5.6	14
105	An Economic Evaluation of Neonatal Screening for Inborn Errors of Metabolism Using Tandem Mass Spectrometry in Thailand. PLoS ONE, 2015, 10, e0134782.	2.5	16
106	Meta-analysis of two Chinese populations identifies an autoimmune disease risk allele in 22q11.21 as associated with systemic lupus erythematosus. Arthritis Research and Therapy, 2015, 17, 67.	3.5	6
107	A Frameshift Mutation in <i>PEN-2</i> Causes Familial Comedones Syndrome. Dermatology, 2015, 231, 77-81.	2.1	10
108	HLAreporter: a tool for HLA typing from next generation sequencing data. Genome Medicine, 2015, 7, 25.	8.2	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. Human Molecular Genetics, 2015, 24, 274-284.	2.9	35
110	Novel AQP2 mutation causing congenital nephrogenic diabetes insipidus: challenges in management during infancy. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 193-7.	0.9	8
111	Two novel mutations including a large deletion of theSLC4A11gene causing autosomal recessive hereditary endothelial dystrophy. British Journal of Ophthalmology, 2014, 98, 1460-1462.	3.9	9
112	Absent expression of the osteoblast-specific maternally imprinted genes,DLX5andDLX6,causes split hand/split foot malformation type I. Journal of Medical Genetics, 2014, 51, 817-823.	3.2	21
113	Three SNPs in chromosome 11q23.3 are independently associated with systemic lupus erythematosus in Asians. Human Molecular Genetics, 2014, 23, 524-533.	2.9	29
114	ZRS 406A>G mutation in patients with tibial hypoplasia, polydactyly and triphalangeal first fingers. Journal of Human Genetics, 2014, 59, 467-470.	2.3	20
115	Genetics and genomics in <scp>T</scp>hailand: challenges and opportunities. Molecular Genetics & Genomic Medicine, 2014, 2, 210-216.	1.2	18
116	In vitro Correction of a Novel Splicing Alteration in the BTK Gene by Using Antisense Morpholino Oligonucleotides. Archivum Immunologiae Et Therapiae Experimentalis, 2014, 62, 431-436.	2.3	2
117	Germline and SomaticDICER1Mutations in a Pituitary Blastoma Causing Infantile-Onset Cushing's Disease. Journal of Clinical Endocrinology and Metabolism, 2014, 99, E1487-E1492.	3.6	73
118	Expanding Phenotypic Spectrum of Familial Comedones. Dermatology, 2014, 228, 215-219.	2.1	9
119	Whole Genome and Exome Sequencing of Monozygotic Twins with Trisomy 21, Discordant for a Congenital Heart Defect and Epilepsy. PLoS ONE, 2014, 9, e100191.	2.5	42
120	Disorders with similar clinical phenotypes reveal underlying genetic interaction: SATB2 acts as an activator of the UPF3B gene. Human Genetics, 2013, 132, 1383-1393.	3.8	24
121	Functional characterization of novel variants in the CETP promoter and the LIPC gene in subjects with hyperalphalipoproteinemia. Clinica Chimica Acta, 2013, 416, 92-95.	1.1	2
122	Meta-analysis Followed by Replication Identifies Loci in or near CDKN1B, TET3, CD80, DRAM1, and ARID5B as Associated with Systemic Lupus Erythematosus in Asians. American Journal of Human Genetics, 2013, 92, 41-51.	6.2	184
123	A newly identified locus for benign adult familial myoclonic epilepsy on chromosome 3q26.32-3q28. European Journal of Human Genetics, 2013, 21, 225-228.	2.8	55
124	DcR3 Mutations in Patients with Juvenile-onset Systemic Lupus Erythematosus Lead to Enhanced Lymphocyte Proliferation. Journal of Rheumatology, 2013, 40, 1316-1326.	2.0	5
125	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	1.2	148
126	Novel <scp>CTSK</scp> mutation resulting in an entire exon 2 skipping in a <scp>T</scp>hai girl with pycnodysostosis. Pediatrics International, 2013, 55, 651-655.	0.5	7

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127	A common and two novel GBA mutations in Thai patients with Gaucher disease. Journal of Human Genetics, 2013, 58, 594-599.	2.3	17
128	FOXE1 mutations in Thai patients with oral clefts. Genetical Research, 2013, 95, 133-137.	0.9	7
129	FGFR1 and FGFR2 Mutations in Pfeiffer Syndrome. Journal of Craniofacial Surgery, 2013, 24, 150-152.	0.7	31
130	Clinical and molecular findings in Thai patients with isolated methylmalonic acidemia. Molecular Genetics and Metabolism, 2012, 106, 424-429.	1.1	29
131	PDGFRa mutations in humans with isolated cleft palate. European Journal of Human Genetics, 2012, 20, 1058-1062.	2.8	55
132	Two novel CTNS mutations in cystinosis patients in Thailand. Gene, 2012, 499, 323-325.	2.2	14
133	Novel CYP11B2 mutation causing aldosterone synthase (P450c11AS) deficiency. European Journal of Pediatrics, 2012, 171, 1559-1562.	2.7	12
134	Primary hyperoxaluria type 1 and brachydactyly mental retardation syndrome caused by a novel mutation in AGXT and a terminal deletion of chromosome 2. American Journal of Medical Genetics, Part A, 2012, 158A, 2124-2130.	1.2	12
135	Pharmacogenetic screening of carbamazepine-induced severe cutaneous allergic reactions. Journal of Clinical Neuroscience, 2011, 18, 1289-1294.	1.5	41
136	Two siblings with a novel nonsense mutation, p.R50X, in the vitamin D receptor gene. Endocrine, 2011, 40, 62-66.	2.3	8
137	PTPRF is disrupted in a patient with syndromic amastia. BMC Medical Genetics, 2011, 12, 46.	2.1	11
138	ELF1 is associated with systemic lupus erythematosus in Asian populations. Human Molecular Genetics, 2011, 20, 601-607.	2.9	78
139	A novel p.E276K IDUA mutation decreasing Î±-L-iduronidase activity causes mucopolysaccharidosis type I. Molecular Vision, 2011, 17, 456-60.	1.1	7
140	Concurrent bilateral pheochromocytoma and thoracic paraganglioma during pregnancy. Endocrine, 2010, 37, 261-264.	2.3	8
141	HLAâ€B* 1502 screening: Time to clinical practice. Epilepsia, 2010, 51, 936-938.	5.1	28
142	Functional Characterization of Vasopressin Receptor 2 Mutations Causing Partial and Complete Congenital Nephrogenic Diabetes Insipidus in Thai Families. Hormone Research in Paediatrics, 2010, 73, 349-354.	1.8	14
143	p.D645E of Acid Î±-Glucosidase Is the Most Common Mutation in Thai Patients with Infantile-Onset Pompe Disease. Genetic Testing and Molecular Biomarkers, 2010, 14, 835-837.	0.7	11
144	Three novel mutations of the IRF6 gene with one associated with an unusual feature in Van der Woude syndrome. American Journal of Medical Genetics, Part A, 2009, 149A, 2489-2492.	1.2	15

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145	Bilateral pheochromocytoma during the postpartum period. Archives of Gynecology and Obstetrics, 2009, 280, 1055-1058.	1.7	17
146	Expanding clinical spectrum of nonâ€autoimmune hyperthyroidism due to an activating germline mutation, p.M453T, in the thyrotropin receptor gene. Clinical Endocrinology, 2009, 70, 623-628.	2.4	21
147	Novel mutations in the STK11 gene in Thai patients withPeutz-Jeghers syndrome. World Journal of Gastroenterology, 2009, 15, 5364.	3.3	7
148	Identification of two novel aquaporin-2 mutations in a Thai girl with congenital nephrogenic diabetes insipidus. Endocrine, 2008, 33, 210-214.	2.3	7
149	Carbamazepine and phenytoin induced Stevensâ€Johnson syndrome is associated with HLAâ€B*1502 allele in Thai population. Epilepsia, 2008, 49, 2087-2091.	5.1	413
150	Risk factors associated with the occurrence of frontoethmoidal encephalomeningocele. European Journal of Paediatric Neurology, 2008, 12, 102-107.	1.6	25
151	Identification of mutations in the SRD5A2 gene in Thai patients with male pseudohermaphroditism. Fertility and Sterility, 2008, 90, 2015.e11-2015.e15.	1.0	25
152	ASSOCIATION OF CYTOKINE-RELATED GENE EXPRESSION WITH DENGUE INFECTION SEVERITY. Pediatrics, 2008, 121, S132.1-S132.	2.1	2
153	A Novel Germline Mutation, IVS4+1G>A, of the POU1F1 Gene Underlying Combined Pituitary Hormone Deficiency. Hormone Research in Paediatrics, 2008, 69, 60-64.	1.8	17
154	Expression of Mammaglobins A and B in Nasal Polyps is Similar in Patients with and without Allergic Rhinitis. American Journal of Rhinology & Allergy, 2008, 22, 135-138.	2.2	2
155	Two novel EBP mutations in Conradi-HÃ¼nermann-Happle syndrome. European Journal of Dermatology, 2008, 18, 391-3.	0.6	14
156	An exceptionally low percentage of Thai expectant mothers and medical personnel with folic acid knowledge and peri-conceptional consumption urges an urgent education program and/or food fortification. International Journal of Food Sciences and Nutrition, 2007, 58, 297-303.	2.8	8
157	Heterozygous nonsense mutationSATB2 associated with cleft palate, osteoporosis, and cognitive defects. Human Mutation, 2007, 28, 732-738.	2.5	120
158	Prenatal exclusion of subtelomeric deletion 1p by fluorescent in situ hybridization. Archives of Gynecology and Obstetrics, 2007, 275, 237-240.	1.7	0
159	MSX1 mutations contribute to nonsyndromic cleft lip in a Thai population. Journal of Human Genetics, 2006, 51, 671-676.	2.3	55
160	No detectable genomic aberrations by BAC array CGH in Kabuki make-up syndrome patients. American Journal of Medical Genetics, Part A, 2006, 140A, 291-293.	1.2	19
161	Tetralogy of Fallot with absent pulmonary valve in a de novo derivative chromosome 9 with duplication of 9p13â€‰â†’â€‰9pter and deletion of 9q34.3. American Journal of Medical Genetics, Part A, 2006, 140A, 1981-1987.	1.2	7
162	A Novel Mutation in EFNB1, Probably with a Dominant Negative Effect, Underlying Craniofrontonasal Syndrome. Cleft Palate-Craniofacial Journal, 2006, 43, 152-154.	0.9	27

#	Article	IF	Citations
163	A Girl with a Novel Splice Site Mutation in VDR Supports the Role of a Ligand-Independent VDR Function on Hair Cycling. Hormone Research in Paediatrics, 2006, 66, 273-276.	1.8	16
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