Vorasuk Shotelersuk

List of Publications by Year
in descending order

195 papers	5,626 citations	117625 34 h-index	95266 68 g-index
197 all docs	197 docs citations	197 times ranked	7498 citing authors

#	Article	IF	Citations
1	Altered Trafficking of Lysosomal Proteins in Hermansky-Pudlak Syndrome Due to Mutations in the Î²3A Subunit of the AP-3 Adaptor. Molecular Cell, 1999, 3, 11-21.	9.7	631
2	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
3	Genetic Defects and Clinical Characteristics of Patients with a Form of Oculocutaneous Albinism (Hermanskyâ€“Pudlak Syndrome). New England Journal of Medicine, 1998, 338, 1258-1265.	27.0	282
4	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
5	Pulmonary Function and High-Resolution CT Findings in Patients With an Inherited Form of Pulmonary Fibrosis, Hermansky-Pudlak Syndrome, Due to Mutations in HPS-1. Chest, 2000, 117, 129-136.	0.8	169
6	CTNS Mutations in an American-Based Population of Cystinosis Patients. American Journal of Human Genetics, 1998, 63, 1352-1362.	6.2	153
7	MLL2 and KDM6A mutations in patients with Kabuki syndrome. American Journal of Medical Genetics, Part A, 2013, 161, 2234-2243.	1.2	148
8	Heterozygous nonsense mutationSATB2 associated with cleft palate, osteoporosis, and cognitive defects. Human Mutation, 2007, 28, 732-738.	2.5	120
9	The Genomic Region Encompassing the Nephropathic Cystinosis Gene (CTNS): Complete Sequencing of a 200-kb Segment and Discovery of a Novel Gene within the Common Cystinosis-Causing Deletion. Genome Research, 2000, 10, 165-173.	5.5	118
10	MBTPS2 mutations cause defective regulated intramembrane proteolysis in X-linked osteogenesis imperfecta. Nature Communications, 2016, 7, 11920.	12.8	112
11	A new variant of Hermansky-Pudlak syndrome due to mutations in a gene responsible for vesicle formation. American Journal of Medicine, 2000, 108, 423-427.	1.5	111
12	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
13	Ocular Nonnephropathic Cystinosis: Clinical, Biochemical, and Molecular Correlations. Pediatric Research, 2000, 47, 17-17.	2.3	103
14	TTTCA repeat insertions in an intron of YEATS2 in benign adult familial myoclonic epilepsy type 4. Brain, 2019, 142, 3360-3366.	7.6	83
15	Hermansky-Pudlak Syndrome: Models for Intracellular Vesicle Formation. Molecular Genetics and Metabolism, 1998, 65, 85-96.	1.1	81
16	Identification and Detection of the Common 65-kb Deletion Breakpoint in the Nephropathic Cystinosis Gene (CTNS). Molecular Genetics and Metabolism, 1999, 66, 111-116.	1.1	81
17	ELF1 is associated with systemic lupus erythematosus in Asian populations. Human Molecular Genetics, 2011, 20, 601-607.	2.9	78
18	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

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19	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
20	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
21	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
22	Evidence for Locus Heterogeneity in Puerto Ricans with Hermansky-Pudlak Syndrome. American Journal of Human Genetics, 1997, 61, 1088-1094.	6.2	67
23	CTNS mutations in patients with cystinosis. Human Mutation, 1999, 14, 454-458.	2.5	63
24	HLAreporter: a tool for HLA typing from next generation sequencing data. Genome Medicine, 2015, 7, 25.	8.2	62
25	MSX1 mutations contribute to nonsyndromic cleft lip in a Thai population. Journal of Human Genetics, 2006, 51, 671-676.	2.3	55
26	PDGFRa mutations in humans with isolated cleft palate. European Journal of Human Genetics, 2012, 20, 1058-1062.	2.8	55
27	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
28	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
29	Three New Mutations in a Gene Causing Hermanskyâ€“Pudlak Syndrome: Clinical Correlations. Molecular Genetics and Metabolism, 1998, 64, 99-107.	1.1	54
30	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
31	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
32	Pharmacogenetic screening of carbamazepine-induced severe cutaneous allergic reactions. Journal of Clinical Neuroscience, 2011, 18, 1289-1294.	1.5	41
33	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
34	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
35	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
36	Generation and characterization of HLA-universal platelets derived from induced pluripotent stem cells. Scientific Reports, 2020, 10, 8472.	3.3	35

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37	CTNS Mutations in African American Patients with Cystinosis. Molecular Genetics and Metabolism, 2001, 74, 332-337.	1.1	33
38	On the reported 8p22-p23.1 duplication in Kabuki make-up syndrome (KMS) and its absence in patients with typical KMS. American Journal of Medical Genetics Part A, 2004, 128A, 170-172.	2.4	32
39	Normal brain myelination in a patient homozygous for a mutation that encodes a severely truncated methionine adenosyltransferase I/III. American Journal of Medical Genetics Part A, 1998, 75, 395-400.	2.4	31
40	FGFR1 and FGFR2 Mutations in Pfeiffer Syndrome. Journal of Craniofacial Surgery, 2013, 24, 150-152.	0.7	31
41	Distinct craniofacial-skeletal-dermatological dysplasia in a patient with W290C mutation inFGFR2. American Journal of Medical Genetics Part A, 2002, 113, 4-8.	2.4	30
42	Clinical and molecular findings in Thai patients with isolated methylmalonic acidemia. Molecular Genetics and Metabolism, 2012, 106, 424-429.	1.1	29
43	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
44	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
45	HLAâ€B* 1502 screening: Time to clinical practice. Epilepsia, 2010, 51, 936-938.	5.1	28
46	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
47	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
48	Kabuki syndrome: Report of six Thai children and further phenotypic and genetic delineation. American Journal of Medical Genetics Part A, 2002, 110, 384-390.	2.4	26
49	Risk factors associated with the occurrence of frontoethmoidal encephalomeningocele. European Journal of Paediatric Neurology, 2008, 12, 102-107.	1.6	25
50	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
51	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
52	Genotypeâ€“phenotype correlation and expansion of orodental anomalies in LTBP3-related disorders. Molecular Genetics and Genomics, 2019, 294, 773-787.	2.1	24
53	The Thai reference exome (Tâ€REx) variant database. Clinical Genetics, 2021, 100, 703-712.	2.0	24
54	Novel mutations in a Thai patient with methylmalonic acidemia. Molecular Genetics and Metabolism, 2003, 79, 300-302.	1.1	21

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55	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
56	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
57	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
58	Prenatal diagnosis of Pompe disease by electron microscopy. Archives of Gynecology and Obstetrics, 2005, 271, 260-262.	1.7	20
59	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
60	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
61	Compromised alveolar bone cells in a patient with dentinogenesis imperfecta caused by DSPP mutation. Clinical Oral Investigations, 2019, 23, 303-313.	3.0	19
62	Discordance of oral-facial-digital syndrome type 1 in monozygotic twin girls. , 1999, 86, 269-273.		18
63	FGFR2 Mutations among Thai Children with Crouzon and Apert Syndromes. Journal of Craniofacial Surgery, 2003, 14, 101-104.	0.7	18
64	Genetics and genomics in <scp>T</scp>hailand: challenges and opportunities. Molecular Genetics & Genomic Medicine, 2014, 2, 210-216.	1.2	18
65	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
66	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
67	Bilateral pheochromocytoma during the postpartum period. Archives of Gynecology and Obstetrics, 2009, 280, 1055-1058.	1.7	17
68	A common and two novel GBA mutations in Thai patients with Gaucher disease. Journal of Human Genetics, 2013, 58, 594-599.	2.3	17
69	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
70	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
71	Two novel compound heterozygous BMP1 mutations in a patient with osteogenesis imperfecta: a case report. BMC Medical Genetics, 2017, 18, 25.	2.1	16
72	The phenotypic and mutational spectrum of Thai female patients with ornithine transcarbamylase deficiency. Gene, 2018, 679, 377-381.	2.2	16

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73	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
74	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
75	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
76	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
77	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
78	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
79	A novel mutation, 1234del(C), of the IRF6 in a Thai family with Van der Woude syndrome. International Journal of Molecular Medicine, 2003, 11, 505-7.	4.0	15
80	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
81	Two novel CTNS mutations in cystinosis patients in Thailand. Gene, 2012, 499, 323-325.	2.2	14
82	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
83	Massive parallel sequencing as a new diagnostic approach for phenylketonuria and tetrahydrobiopterin-deficiency in Thailand. BMC Medical Genetics, 2017, 18, 102.	2.1	14
84	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
85	Precision medicine in Thailand. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 245-253.	1.6	14
86	Founder effect of the TTTCA repeat insertions in SAMD12 causing BAFME1. European Journal of Human Genetics, 2021, 29, 343-348.	2.8	14
87	Two novel EBP mutations in Conradi-HÃ¼nermann-Happle syndrome. European Journal of Dermatology, 2008, 18, 391-3.	0.6	14
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	Amelogenesis imperfecta: A novel FAM83H mutation and characteristics of periodontal ligament cells. Oral Diseases, 2018, 24, 1522-1531.	3.0	13
90	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

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91	Novel CYP11B2 mutation causing aldosterone synthase (P450c11AS) deficiency. European Journal of Pediatrics, 2012, 171, 1559-1562.	2.7	12
92	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
93	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
94	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
95	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
96	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
97	PTPRF is disrupted in a patient with syndromic amastia. BMC Medical Genetics, 2011, 12, 46.	2.1	11
98	Coleâ€Carpenter syndrome in a patient from Thailand. American Journal of Medical Genetics, Part A, 2018, 176, 1706-1710.	1.2	11
99	MBTPS2, a membrane bound protease, underlying several distinct skin and bone disorders. Journal of Translational Medicine, 2021, 19, 114.	4.4	11
100	A Frameshift Mutation in <i>PEN-2</i> Causes Familial Comedones Syndrome. Dermatology, 2015, 231, 77-81.	2.1	10
101	A Pathogenic Variant inALPK3Is Associated With an Autosomal Dominant Adult-onset Hypertrophic Cardiomyopathy. Circulation Genomic and Precision Medicine, 2020, 13, e003127.	3.6	10
102	PTEN c.511C>T Nonsense Mutation in a BRRS Family Disrupts a Potential Exonic Splicing Enhancer and Causes Exon Skipping. Japanese Journal of Clinical Oncology, 2006, 36, 814-821.	1.3	9
103	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
104	Expanding Phenotypic Spectrum of Familial Comedones. Dermatology, 2014, 228, 215-219.	2.1	9
105	Splicing analysis of CYP11B1 mutation in a family affected with 11Î²-hydroxylase deficiency: case report. BMC Endocrine Disorders, 2016, 16, 37.	2.2	9
106	Human asparagine synthetase associates with the mitotic spindle. Biology Open, 2018, 7, .	1.2	9
107	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
108	Phenotypic and Genotypic Features of Thai Patients With Nonsyndromic Tooth Agenesis and WNT10A Variants. Frontiers in Physiology, 2020, 11, 573214.	2.8	9

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109	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
110	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
111	Concurrent bilateral pheochromocytoma and thoracic paraganglioma during pregnancy. Endocrine, 2010, 37, 261-264.	2.3	8
112	Two siblings with a novel nonsense mutation, p.R50X, in the vitamin D receptor gene. Endocrine, 2011, 40, 62-66.	2.3	8
113	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
114	Novel Mutations, Including a Large Deletion in theARSBGene, Causing Mucopolysaccharidosis Type VI. Genetic Testing and Molecular Biomarkers, 2017, 21, 58-62.	0.7	8
115	Female-restricted syndromic intellectual disability in a patient from Thailand. , 2019, 179, 758-761.		8
116	A family with homozygous and heterozygous p.Gly337Ser mutations in COL1A2. European Journal of Medical Genetics, 2020, 63, 103896.	1.3	8
117	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
118	Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Gene, 2020, 749, 144709.	2.2	8
119	Cleidocranial dysplasia and novel RUNX2 variants: dental, craniofacial, and osseous manifestations. Journal of Applied Oral Science, 0, 30, .	1.8	8
120	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
121	Identification of two novel aquaporin-2 mutations in a Thai girl with congenital nephrogenic diabetes insipidus. Endocrine, 2008, 33, 210-214.	2.3	7
122	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
123	FOXE1 mutations in Thai patients with oral clefts. Genetical Research, 2013, 95, 133-137.	0.9	7
124	Widespread and debilitating hemangiomas in a patient with enchondromatosis and D-2-hydroxyglutaric aciduria. Skeletal Radiology, 2018, 47, 1577-1582.	2.0	7
125	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
126	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

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127	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
128	Congenital myasthenic syndromes in the Thai population: Clinical findings and novel mutations. Neuromuscular Disorders, 2020, 30, 851-858.	0.6	7
129	Tooth ultrastructure of a novel COL1A2 mutation expanding its genotypic and phenotypic spectra. Oral Diseases, 2021, 27, 1257-1267.	3.0	7
130	Novel mutations in the STK11 gene in Thai patients withPeutz-Jeghers syndrome. World Journal of Gastroenterology, 2009, 15, 5364.	3.3	7
131	Whole exome sequencing for diagnosis of hereditary thrombocytopenia. Medicine (United States), 2020, 99, e23275.	1.0	7
132	A novel p.E276K IDUA mutation decreasing Î±-L-iduronidase activity causes mucopolysaccharidosis type I. Molecular Vision, 2011, 17, 456-60.	1.1	7
133	ASA E382K disrupts a potential exonic splicing enhancer and causes exon skipping, but missense mutations in ASA are not associated with ESEs. International Journal of Molecular Medicine, 2004, 14, 683-9.	4.0	7
134	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
135	Epidemiology of cleft lip with or without cleft palate in Thais. Asian Biomedicine, 2017, 10, 335-338.	0.3	6
136	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
137	Expanding phenotypic and mutational spectra of mitochondrial HMG-CoA synthase deficiency. European Journal of Medical Genetics, 2020, 63, 104086.	1.3	6
138	Reduced ELANE and SLPI expression compromises dental pulp cell activity. Cell Proliferation, 2021, 54, e13132.	5.3	6
139	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
140	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
141	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
142	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
143	Novel de novo mutation substantiates ATP6V0C as a gene causing epilepsy with intellectual disability. Brain and Development, 2021, 43, 490-494.	1.1	5
144	Prenatal Sonographic Features of CHARGE Syndrome. Diagnostics, 2021, 11, 415.	2.6	5

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145	Actionable secondary findings in the 73 ACMG-recommended genes in 1559 Thai exomes. Journal of Human Genetics, 2021, , .	2.3	5
146	A Novel Germline Mutation, 1793delG, of the MEN1 Gene Underlying Multiple Endocrine Neoplasia Type 1. Japanese Journal of Clinical Oncology, 2005, 35, 280-282.	1.3	4
147	rs11567842 SNP in SLC13A2 gene associates with hypocitraturia in Thai patients with nephrolithiasis. Genes and Genomics, 2018, 40, 965-972.	1.4	4
148	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
149	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
150	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
151	A novel termination codon mutation of the WAS gene in a Thai family with Wiskott-Aldrich syndrome. International Journal of Molecular Medicine, 2003, 12, 939-41.	4.0	4
152	Genotypic and phenotypic landscapes of 51 pharmacogenes derived from whole-genome sequencing in a Thai population. PLoS ONE, 2022, 17, e0263621.	2.5	4
153	Postnatal growth failure, microcephaly, mental retardation, cataracts, large joint contractures, osteoporosis, cortical dysplasia, and cerebellar atrophy. American Journal of Medical Genetics Part A, 2003, 116A, 164-169.	2.4	3
154	A novel PCCB mutation in a Thai patient with propionic acidemia identified by exome sequencing. Human Genome Variation, 2015, 2, 15033.	0.7	3
155	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
156	Novel mutations in Thai patients with glanzmann thrombasthenia. European Journal of Haematology, 2017, 99, 520-524.	2.2	3
157	A Novel <i>GNAS</i> Mutation Causing Isolated Infantile Cushingâ€™s Syndrome. Hormone Research in Paediatrics, 2019, 92, 196-202.	1.8	3
158	Clinical and molecular characteristics of Thai patients with ELANE-related neutropaenia. Journal of Clinical Pathology, 2020, , jclinpath-2020-207139.	2.0	3
159	Whole-Exome Sequencing Solved over 2-Decade Kidney Disease Enigma. Nephron, 2021, 145, 311-316.	1.8	3
160	Ageâ€related dental phenotypes and tooth characteristics of FAM83Hâ€associated hypocalcified amelogenesis imperfecta. Oral Diseases, 2022, 28, 734-744.	3.0	3
161	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
162	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

#	Article	IF	Citations
163	A novel nonsense mutation, E150X, in the SOX9 gene underlying campomelic dysplasia. Genetics and Molecular Biology, 2006, 29, 617-620.	1.3	3
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