Pei-Lung Chen

List of Publications by Year
in descending order

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90	3,729	236925 25	133252 59
papers	citations	h-index	g-index

96	96	96	5832
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Investigating DYT1 in a Taiwanese dystonia cohort. Journal of the Formosan Medical Association, 2022, 121, 375-380.	1.7	3
2	Clinical manifestations and genetic characteristics in the Taiwan thoracic aortic aneurysm and dissection cohort - a prospective cohort study. Journal of the Formosan Medical Association, 2022, 121, 1093-1101.	1.7	5
3	Cochlear implantation in LEOPARD syndrome: Our experience with three patients. Clinical Otolaryngology, 2022, 47, 341-346.	1.2	0
4	A Clinical and Integrated Genetic Study of Isolated and Combined Dystonia in Taiwan. Journal of Molecular Diagnostics, 2022, 24, 262-273.	2.8	11
5	Genetic analysis of a family presenting with coexisting cerebral cavernous malformations and polycystic kidney disease. Journal of the Formosan Medical Association, 2022, , .	1.7	0
6	PREDICTED PROTEIN STRUCTURE VARIATIONS INDICATE THE CLINICAL PRESENTATION OF CYP4V2-RELATED BIETTI CRYSTALLINE DYSTROPHY. Retina, 2022, 42, 797-806.	1.7	4
7	Risk factors related to age at diagnosis of pancreatic cancer: a retrospective cohort pilot study. BMC Gastroenterology, 2022, 22, 243.	2.0	4
8	Cochlear Implantation Outcomes in Patients with Auditory Neuropathy Spectrum Disorder of Genetic and Non-Genetic Etiologies: A Multicenter Study. Biomedicines, 2022, 10, 1523.	3.2	9
9	Genotypes Influence Clinical Progression in EYS-Associated Retinitis Pigmentosa. Translational Vision Science and Technology, 2022, 11, 6.	2.2	2
10	Next-generation sequencing and bioinformatics to identify genetic causes of malignant hyperthermia. Journal of the Formosan Medical Association, 2021, 120, 883-892.	1.7	6
11	Genetic study of<scp>youngâ€onset</scp>dementia using targeted gene panel sequencing in Taiwan. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2021, 186, 67-76.	1.7	4
12	Metabolic Syndromes as Important Comorbidities in Patients of Inherited Retinal Degenerations: Experiences from the Nationwide Health Database and a Large Hospital-Based Cohort. International Journal of Environmental Research and Public Health, 2021, 18, 2065.	2.6	1
13	Genetic characteristics and epidemiology of inherited retinal degeneration in Taiwan. Npj Genomic Medicine, 2021, 6, 16.	3.8	36
14	Rare manifestations and malignancies in tuberous sclerosis complex: findings from the TuberOus SClerosis registry to increAse disease awareness (TOSCA). Orphanet Journal of Rare Diseases, 2021, 16, 301.	2.7	15
15	Hearing Impairment with Monoallelic GJB2 Variants. Journal of Molecular Diagnostics, 2021, 23, 1279-1291.	2.8	10
16	Genetic Spectrum and Characteristics of Hereditary Optic Neuropathy in Taiwan. Genes, 2021, 12, 1378.	2.4	4
17	The Clinical Contribution of Full-Field Electroretinography and 8-Year Experiences of Application in a Tertiary Medical Center. Journal of Personalized Medicine, 2021, 11, 1022.	2.5	1
18	Hereditary Hearing Impairment with Cutaneous Abnormalities. Genes, 2021, 12, 43.	2.4	3

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19	Evaluation of using WGS/WES to characterize ACMG actionable genes in genetic testing reports. , 2021, , .		0
20	Hearing Features and Cochlear Implantation Outcomes in Patients With Pathogenic MYO15A Variants. Ear and Hearing, 2021, Publish Ahead of Print, .	2.1	1
21	Prediction Model for Audiological Outcomes in Patients With GJB2 Mutations. Ear and Hearing, 2020, 41, 143-149.	2.1	16
22	Genotypes Predispose Phenotypesâ€"Clinical Features and Genetic Spectrum of ABCA4-Associated Retinal Dystrophies. Genes, 2020, 11, 1421.	2.4	8
23	Natural clusters of tuberous sclerosis complex (TSC)-associated neuropsychiatric disorders (TAND): new findings from the TOSCA TAND research project. Journal of Neurodevelopmental Disorders, 2020, 12, 24.	3.1	16
24	An integrative approach for pediatric auditory neuropathy spectrum disorders: revisiting etiologies and exploring the prognostic utility of auditory steady-state response. Scientific Reports, 2020, 10, 9816.	3.3	7
25	Genotype-phenotype correlations of adult-onset PLA2G6-associated Neurodegeneration: case series and literature review. BMC Neurology, 2020, 20, 101.	1.8	29
26	The efficacy and safety of topical rapamycinâ€"calcitriol for facial angiofibromas in patients with tuberous sclerosis complex: a prospective, doubleâ€blind, randomized clinical trial. British Journal of Dermatology, 2020, 183, 655-663.	1.5	14
27	Cerebral Microbleeds in Autosomal Dominant Polycystic Kidney Disease. Journal of Stroke, 2020, 22, 153-156.	3.2	3
28	Serum levels of insulin-like growth factor 1 are negatively associated with log transformation of thyroid-stimulating hormone in Gravesâ€™ disease patients with hyperthyroidism or subjects with euthyroidism. Medicine (United States), 2019, 98, e14862.	1.0	6
29	Plectin Mutations in Progressive Familial Intrahepatic Cholestasis. Hepatology, 2019, 70, 2221-2224.	7.3	12
30	Genetic Epidemiology and Clinical Features of Hereditary Hearing Impairment in the Taiwanese Population. Genes, 2019, 10, 772.	2.4	31
31	Distinctive genetic variation of longâ€segment Hirschsprung's disease in Taiwan. Neurogastroenterology and Motility, 2019, 31, e13665.	3.0	7
32	HSD3B1 gene polymorphism and female pattern hair loss in women with polycystic ovary syndrome. Journal of the Formosan Medical Association, 2019, 118, 1225-1231.	1.7	9
33	A clinical and genetic study of earlyâ€onset and familial parkinsonism in taiwan: An integrated approach combining gene dosage analysis and nextâ€generation sequencing. Movement Disorders, 2019, 34, 506-515.	3.9	71
34	P53 ICE CRIM mouse: a tool to generate mutant allelic series in somatic cells and germ lines for cancer studies. FASEB Journal, 2019, 33, 5571-5584.	0.5	1
35	Targeted Next-Generation Sequencing Facilitates Genetic Diagnosis and Provides Novel Pathogenetic Insights into Deafness with Enlarged Vestibular Aqueduct. Journal of Molecular Diagnostics, 2019, 21, 138-148.	2.8	9
36	Blood group genotyping goes next generation: featuring <scp>ABO</scp>,<scp> RH</scp> and <scp>MNS</scp>. ISBT Science Series, 2018, 13, 290-297.	1.1	2

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37	Comprehensive human leukocyte antigen genotyping of patients with type 1 diabetes mellitus in Taiwan. Pediatric Diabetes, 2018, 19, 699-706.	2.9	7
38	A 1204-single nucleotide polymorphism and insertionâ€“deletion polymorphism panel for massively parallel sequencing analysis of DNA mixtures. Forensic Science International: Genetics, 2018, 32, 94-101.	3.1	38
39	Using Ion Torrent sequencing to study genetic mutation profiles of fatal thyroid cancers. Journal of the Formosan Medical Association, 2018, 117, 488-496.	1.7	3
40	Serum levels of fetuin-A are negatively associated with log transformation levels of thyroid-stimulating hormone in patients with hyperthyroidism or euthyroidism. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.6	4
41	Unique spectra of deafness-associated mutations in Mongolians provide insights into the genetic relationships among Eurasian populations. PLoS ONE, 2018, 13, e0209797.	2.5	12
42	Overview of the current status of familial hypercholesterolaemia care in over 60 countries - The EAS Familial Hypercholesterolaemia Studies Collaboration (FHSC). Atherosclerosis, 2018, 277, 234-255.	0.8	163
43	Identification of a novel LDLR disease-causing variant using capture-based next-generation sequencing screening of familial hypercholesterolemia patients in Taiwan. Atherosclerosis, 2018, 277, 440-447.	0.8	11
44	ABO genotyping with nextâ€generation sequencing to resolve heterogeneity in donors with serology discrepancies. Transfusion, 2018, 58, 2232-2242.	1.6	37
45	The Lupusâ€Associated FcÎ³ Receptor IIbâ€“I232T Polymorphism Results in Impairment in the Negative Selection of Lowâ€Affinity Germinal Center B Cells Via câ€Abl in Mice. Arthritis and Rheumatology, 2018, 70, 1866-1878.	5.6	5
46	Thyrotropin receptor antibodies and a genetic hint in antithyroid drug-induced adverse drug reactions. Expert Opinion on Drug Safety, 2018, 17, 775-784.	2.4	5
47	Oncogenic Function of a KIF5B-MET Fusion Variant in Non-Small Cell Lung Cancer. Neoplasia, 2018, 20, 838-847.	5.3	25
48	Early measurement of IL-10 predicts the outcomes of patients with acute respiratory distress syndrome receiving extracorporeal membrane oxygenation. Scientific Reports, 2017, 7, 1021.	3.3	27
49	Identification of a novel HLAâ€B allele, HLAâ€B*40:238, in a Taiwanese individual. Hla, 2017, 90, 252-253.	0.6	2
50	A novel missense variant in the nuclear localization signal of POU4F3 causes autosomal dominant non-syndromic hearing loss. Scientific Reports, 2017, 7, 7551.	3.3	18
51	Intrafamilial phenotypic heterogeneity in a Taiwanese family with a MAPT p.R5H mutation: a case report and literature review. BMC Neurology, 2017, 17, 186.	1.8	12
52	Identification of VPS35 p.D620N mutation-related Parkinsonâ€™s disease in a Taiwanese family with successful bilateral subthalamic nucleus deep brain stimulation: a case report and literature review. BMC Neurology, 2017, 17, 191.	1.8	20
53	Multiple gene sequencing for risk assessment in patients with early-onset or familial breast cancer. Oncotarget, 2016, 7, 8310-8320.	1.8	83
54	Clinical heterogeneity of LRRK2 p.I2012T mutation. Parkinsonism and Related Disorders, 2016, 33, 36-43.	2.2	17

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55	Neuregulin 3 Knockout Mice Exhibit Behaviors Consistent with Psychotic Disorders. Molecular Neuropsychiatry, 2016, 2, 79-87.	2.9	27
56	KCNN2 polymorphisms and cardiac tachyarrhythmias. Medicine (United States), 2016, 95, e4312.	1.0	11
57	Serum Spot 14 concentration is negatively associated with thyroid-stimulating hormone level. Medicine (United States), 2016, 95, e5036.	1.0	4
58	First step towards precision medicine for antithyroid drug-induced agranulocytosis. Lancet Diabetes and Endocrinology,the, 2016, 4, 473-474.	11.4	1
59	Follicular thyroid carcinoma with NRAS Q61K and GNAS R201H mutations that had a good 131I treatment response. Endocrinology, Diabetes and Metabolism Case Reports, 2016, 2016, 150067.	0.5	3
60	Identification and Functional Studies of Regulatory Variants Responsible for the Association of <i>NRG3</i> with a Delusion Phenotype in Schizophrenia. Molecular Neuropsychiatry, 2015, 1, 36-46.	2.9	14
61	Identifying Children With Poor Cochlear Implantation Outcomes Using Massively Parallel Sequencing. Medicine (United States), 2015, 94, e1073.	1.0	50
62	Long-Term Cochlear Implant Outcomes in Children with GJB2 and SLC26A4 Mutations. PLoS ONE, 2015, 10, e0138575.	2.5	38
63	Functional Variants in DPYSL2 Sequence Increase Risk of Schizophrenia and Suggest a Link to mTOR Signaling. G3: Genes, Genomes, Genetics, 2015, 5, 61-72.	1.8	39
64	Concurrent exome-targeted next-generation sequencing and single nucleotide polymorphism array to identify the causative genetic aberrations of isolated Mayer-Rokitansky-Kuster-Hauser syndrome. Human Reproduction, 2015, 30, 1732-1742.	0.9	19
65	Genetic determinants of antithyroid drug-induced agranulocytosis by human leukocyte antigen genotyping and genome-wide association study. Nature Communications, 2015, 6, 7633.	12.8	93
66	Identification of a novel GATA3 mutation in a deaf Taiwanese family by massively parallel sequencing. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2015, 771, 1-5.	1.0	11
67	An isodicentric X chromosome with gonadal dysgenesis in a lady without prominent somatic features of Turner's syndrome. A case report. Journal of the Formosan Medical Association, 2015, 114, 77-80.	1.7	5
68	Next generation sequencing yields the complete mitochondrial genome of the flathead mullet,Mugil cephaluscryptic species NWP2 (Teleostei: Mugilidae). Mitochondrial DNA, 2014, 27, 1-2.	0.6	40
69	Genetic Analysis of CARD14 in Non-familial Pityriasis Rubra Pilaris: A Case Series. Acta Dermato-Venereologica, 2014, 94, 587-588.	1.3	12
70	STAT3 Mediates Regorafenib-Induced Apoptosis in Hepatocellular Carcinoma. Clinical Cancer Research, 2014, 20, 5768-5776.	7.0	78
71	Predicting HLA genotypes using unphased and flanking single-nucleotide polymorphisms in Han Chinese population. BMC Genomics, 2014, 15, 81.	2.8	12
72	A fault-tolerant method for HLA typing with PacBio data. BMC Bioinformatics, 2014, 15, 296.	2.6	18

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73	Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 243-254.	2.1	1,185
74	Tuberous Sclerosis Complex Surveillance and Management: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatric Neurology, 2013, 49, 255-265.	2.1	693
75	A genome-wide association analysis identifies novel susceptibility loci for coronary arterial lesions in patients with Kawasaki disease. Translational Research, 2013, 161, 513-515.	5.0	12
76	Gene-wide tagging study of the effects of common genetic polymorphisms in the Î± subunits of the GABA_A receptor on epilepsy treatment response. Pharmacogenomics, 2013, 14, 1849-1856.	1.3	19
77	Identification of a novel HLA-B allele,B*07:162, in a Taiwanese individual. Tissue Antigens, 2013, 82, 60-62.	1.0	3
78	Application of Massively Parallel Sequencing to Genetic Diagnosis in Multiplex Families with Idiopathic Sensorineural Hearing Impairment. PLoS ONE, 2013, 8, e57369.	2.5	32
79	Common genetic mutations in the start codon of the SDH subunit D gene among Chinese families with familial head and neck paragangliomas. Oral Oncology, 2012, 48, 125-129.	1.5	9
80	Comprehensive Genotyping in Two Homogeneous Graves' Disease Samples Reveals Major and Novel HLA Association Alleles. PLoS ONE, 2011, 6, e16635.	2.5	60
81	Clinical Implication of the C Allele of the ITPKC Gene SNP rs28493229 in Kawasaki Disease. Pediatric Infectious Disease Journal, 2011, 30, 148-152.	2.0	66
82	Linkage and association on 8p21.2-p21.1 in schizophrenia. , 2011, 156, 188-197.		26
83	Replication of an association of a common variant in the Reelin gene (RELN) with schizophrenia in Ashkenazi Jewish women. Psychiatric Genetics, 2010, 20, 184-186.	1.1	45
84	Human genetics of diabetes mellitus in Taiwan. Frontiers in Bioscience - Landmark, 2009, Volume, 4535.	3.0	3
85	Familiality of Novel Factorial Dimensions of Schizophrenia. Archives of General Psychiatry, 2009, 66, 591.	12.3	71
86	Fine Mapping on Chromosome 10q22-q23 Implicates Neuregulin 3 in Schizophrenia. American Journal of Human Genetics, 2009, 84, 21-34.	6.2	81
87	Family-based association study of cytotoxic T-lymphocyte antigen-4 with susceptibility to Graves' disease in Han population of Taiwan. Genes and Immunity, 2008, 9, 87-92.	4.1	12
88	Linkage of Graves? disease to the human leucocyte antigen region in the Chinese-Han population in Taiwan. Clinical Endocrinology, 2007, 66, 646-651.	2.4	9
89	Translational repression restricts expression of the C. elegans Nanos homolog NOS-2 to the embryonic germline. Developmental Biology, 2006, 292, 244-252.	2.0	72
90	Serum levels of insulin-like growth factor 1 in patients with hyperthyroidism or euthyroidism. Endocrine Abstracts, 0, , .	0.0	0