

# Ene Choo Tan

## List of Publications by Year in descending order

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106  
papers

2,419  
citations

236925

25  
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233421

45  
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108  
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108  
docs citations

108  
times ranked

3865  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical features of a male with a <scp><i>USP9X</i></scp> variant associated with intellectual disability: A case study and review of reported cases. American Journal of Medical Genetics, Part A, 2022, 188, 672-675.	1.2	3
2	Novel phenotypic feature in a patient with a recurrent <scp><i>NOTCH2</i></scp> nonsense mutation. American Journal of Medical Genetics, Part A, 2022, 188, 2135-2138.	1.2	2
3	A novel <i>NSDHL</i> variant in CHILD syndrome with gastrointestinal manifestations and localized skin involvement. Molecular Genetics & Genomic Medicine, 2022, 10, e1848.	1.2	1
4	Genetic landscape of congenital disorders in patients from Southeast Asia: results from sequencing using a gene panel for Mendelian phenotypes. Archives of Disease in Childhood, 2021, 106, 38-43.	1.9	7
5	Self-Improving dystrophic epidermolysis bullosa: First report of clinical, molecular, and genetic characterization of five patients from Southeast Asia. American Journal of Medical Genetics, Part A, 2021, 185, 625-630.	1.2	3
6	Singapore Undiagnosed Disease Program: Genomic Analysis aids Diagnosis and Clinical Management. Archives of Disease in Childhood, 2021, 106, 31-37.	1.9	17
7	Case Report: Mosaicism of a novel nonsense variant in the neurofibromin gene underlies a mosaic generalized NF1 phenotype. F1000Research, 2021, 10, 148.	1.6	0
8	Mindfulness intervention for mild cognitive impairment led to attention-related improvements and neuroplastic changes: Results from a 9-month randomized control trial. Journal of Psychiatric Research, 2021, 135, 203-211.	3.1	26
9	Innate Immune and Neuronal Genetic Markers Are Highly Predictive of Postoperative Pain and Morphine Patient-Controlled Analgesia Requirements in Indian but Not Chinese or Malay Hysterectomy Patients. Pain Medicine, 2021, 22, 2648-2660.	1.9	4
10	Case Report: Mosaicism of a novel nonsense variant in the neurofibromin gene underlies a mosaic generalized NF1 phenotype. F1000Research, 2021, 10, 148.	1.6	0
11	Epidermolysis bullosa with pyloric atresia associated with compound heterozygous <i>ITGB4</i> pathogenic variants: Minimal skin involvement but severe mucocutaneous disease. Pediatric Dermatology, 2021, 38, 908-912.	0.9	5
12	CARD14-associated papulosquamous eruption (CAPE) in a toddler responding to treatment with acitretin. Pediatric Dermatology, 2021, 38, 970-972.	0.9	1
13	Biallelic loss-of-function variants in WDR11 are associated with microcephaly and intellectual disability. European Journal of Human Genetics, 2021, 29, 1663-1668.	2.8	7
14	Mindfulness Awareness Practice (MAP) to Prevent Dementia in Older Adults with Mild Cognitive Impairment: Protocol of a Randomized Controlled Trial and Implementation Outcomes. International Journal of Environmental Research and Public Health, 2021, 18, 10205.	2.6	6
15	MIRAGE Syndrome Caused by a De Novo c.3406G>C (p. Glu1136Gln) Mutation in the SAMD9 Gene Presenting With Neonatal Adrenal Insufficiency and Recurrent Intussusception: A Case Report. Frontiers in Endocrinology, 2021, 12, 742495.	3.5	3
16	Epidermolytic ichthyosis in a child and systematized epidermolytic nevi in the mosaic parent associated with a KRT1 variant. European Journal of Medical Genetics, 2021, 64, 104324.	1.3	3
17	Review and Consensus on Pharmacogenomic Testing in Psychiatry. Pharmacopsychiatry, 2021, 54, 5-17.	3.3	96
18	Microcephaly with a simplified gyral pattern in a child with a de novo <i>TUBA1A</i> variant. American Journal of Medical Genetics, Part A, 2020, 182, 576-578.	1.2	3

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19	Palmoplantar keratoderma, oral involvement, and homozygous CTSC mutation in two brothers from Cambodia. American Journal of Medical Genetics, Part A, 2020, 182, 296-302.	1.2	3
20	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	12.8	43
21	Additional individuals with <scp><i>CHD7</i></scp> variants in Chinese and other southeast Asian patients. American Journal of Medical Genetics, Part A, 2020, 182, 2461-2465.	1.2	0
22	Analysis of SCN9A Gene Variants for Acute and Chronic Postoperative Pain and Morphine Consumption After Total Hysterectomy. Pain Medicine, 2020, 21, 2642-2649.	1.9	2
23	Association of renin-angiotensin-aldosterone system genetic polymorphisms with maternal hypotension during spinal anaesthesia for caesarean delivery: a retrospective cohort study. International Journal of Obstetric Anesthesia, 2020, 44, 3-12.	0.4	3
24	Identification of KMT2D and KDM6A variants by targeted sequencing from patients with Kabuki syndrome and other congenital disorders. Gene, 2020, 731, 144360.	2.2	9
25	Coffin-Siris Syndrome-1: Report of five cases from Asian populations with truncating mutations in the ARID1B gene. Journal of the Neurological Sciences, 2020, 414, 116819.	0.6	5
26	Population genomics in South East Asia captures unexpectedly high carrier frequency for treatable inherited disorders. Genetics in Medicine, 2019, 21, 207-212.	2.4	18
27	&lt;p&gt;Deep sequencing analysis to identify novel and rare variants in pain-related genes in patients with acute postoperative pain and high morphine use&lt;p&gt;. Journal of Pain Research, 2019, Volume 12, 2755-2770.	2.0	4
28	Chromosome 12q24.31 microdeletion and congenital heart disease: a case report and review of the literature. Clinical Dysmorphology, 2019, 28, 53-56.	0.3	0
29	The spectrum of genetic variants and phenotypic features of Southeast Asian patients with Noonan syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e00581.	1.2	8
30	Large expert-curated database for benchmarking document similarity detection in biomedical literature search. Database: the Journal of Biological Databases and Curation, 2019, 2019, .	3.0	15
31	TFAP2A mutation in a child and mother with predominantly ocular anomalies: non-classical presentation of branchio-oculo-facial syndrome. Clinical Dysmorphology, 2019, 28, 215-218.	0.3	7
32	Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. European Journal of Medical Genetics, 2018, 61, 585-595.	1.3	22
33	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	3.2	59
34	Compound heterozygous mutations with novel missenseABCA12mutation in harlequin ichthyosis. BMJ Case Reports, 2018, 2018, bcr-2017-222025.	0.5	7
35	Investigation of variants in estrogen receptor genes and perinatal depression. Neuropsychiatric Disease and Treatment, 2018, Volume 14, 919-925.	2.2	10
36	Piebaldism with multiple café-au-lait-like hyperpigmented macules and inguinal freckling caused by a novel KIT mutation. JAAD Case Reports, 2018, 4, 318-321.	0.8	2

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37	Dataset on gene expression in the elderly after Mindfulness Awareness Practice or Health Education Program. <i>Data in Brief</i> , 2018, 18, 902-912.	1.0	4
38	Intragenic multi-exon deletion in the FBN1 gene in a child with mildly dilated aortic sinus: a retrotransposal event. <i>Journal of Human Genetics</i> , 2017, 62, 711-715.	2.3	9
39	Acute lymphoblastic leukemia in a child with a de novo germline gnb1 mutation. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 550-552.	1.2	17
40	A Novel Interferon Regulatory Factor 6 Mutation in an Asian Family with Van der Woude Syndrome. <i>Cleft Palate-Craniofacial Journal</i> , 2017, 54, 442-445.	0.9	3
41	Correlation of cord blood telomere length with birth weight. <i>BMC Research Notes</i> , 2017, 10, 469.	1.4	24
42	Investigating analgesic and psychological factors associated with risk of postpartum depression development: a case&ndash;control study. <i>Neuropsychiatric Disease and Treatment</i> , 2016, 12, 1333.	2.2	37
43	DICER1 deletion and 14q32 microdeletion syndrome. <i>Clinical Dysmorphology</i> , 2016, 25, 37-40.	0.3	6
44	ARCN1 Mutations Cause a Recognizable Craniofacial Syndrome Due to COPI-Mediated Transport Defects. <i>American Journal of Human Genetics</i> , 2016, 99, 451-459.	6.2	65
45	Ethnicity-dependent influence of innate immune genetic markers on morphine PCA requirements and adverse effects in postoperative pain. <i>Pain</i> , 2016, 157, 2458-2466.	4.2	26
46	Incidentalome from Genomic Sequencing: A Barrier to Personalized Medicine?. <i>EBioMedicine</i> , 2016, 5, 211-216.	6.1	23
47	Common variants of catechol-O-methyltransferase influence patient-controlled analgesia usage and postoperative pain in patients undergoing total hysterectomy. <i>Pharmacogenomics Journal</i> , 2016, 16, 186-192.	2.0	18
48	Case-control study of glucocorticoid receptor and corticotrophin-releasing hormone receptor gene variants and risk of perinatal depression. <i>BMC Pregnancy and Childbirth</i> , 2015, 15, 283.	2.4	13
49	Next-generation sequencing using a pre-designed gene panel for the molecular diagnosis of congenital disorders in pediatric patients. <i>Human Genomics</i> , 2015, 9, 33.	2.9	26
50	Clinical application of next-generation sequencing for Mendelian diseases. <i>Human Genomics</i> , 2015, 9, 10.	2.9	79
51	Tricho-hepato-enteric syndrome (THE-S): two cases and review of the literature. <i>European Journal of Pediatrics</i> , 2015, 174, 1405-1411.	2.7	15
52	Left Ventricular Non-compaction: Is It Genetic?. <i>Pediatric Cardiology</i> , 2015, 36, 1565-1572.	1.3	11
53	Cockayne Syndrome due to a maternally-inherited whole gene deletion of ERCC8 and a paternally-inherited ERCC8 exon 4 deletion. <i>Gene</i> , 2015, 572, 274-278.	2.2	5
54	Massively Parallel Sequencing of Patients with Intellectual Disability, Congenital Anomalies and/or Autism Spectrum Disorders with a Targeted Gene Panel. <i>PLoS ONE</i> , 2014, 9, e93409.	2.5	35

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55	Association of carbamazepine-induced severe cutaneous drug reactions and HLA-B*1502 allele status, and dose and treatment duration in paediatric neurology patients in Singapore. Archives of Disease in Childhood, 2014, 99, 581-584.	1.9	45
56	Chromosome 15q11-q13 copy number gain detected by array-CGH in two cases with a maternal methylation pattern. Molecular Cytogenetics, 2014, 7, 32.	0.9	7
57	Association of premenstrual/menstrual symptoms with perinatal depression and a polymorphic repeat in the polyglutamine tract of the retinoic acid induced 1 gene. Journal of Affective Disorders, 2014, 161, 43-46.	4.1	6
58	De novo 2.3ÂMb microdeletion of 1q32.2 involving the Van der Woude Syndrome locus. Molecular Cytogenetics, 2013, 6, 31.	0.9	16
59	Influence of Mu-Opioid Receptor Variant on Morphine Use and Self-Rated Pain Following Abdominal Hysterectomy. Journal of Pain, 2013, 14, 1045-1052.	1.4	47
60	De novo trisomy 12p in twin girls with different levels of mosaicism. American Journal of Medical Genetics, Part A, 2013, 161, 1702-1705.	1.2	1
61	De novo 3q22.1 q24 deletion associated with multiple congenital anomalies, growth retardation and intellectual disability. Gene, 2013, 517, 82-88.	2.2	4
62	A submicroscopic deletion involving part of the CREBBP gene detected by array-CGH in a patient with Rubinsteinâ€“Taybi syndrome. Gene, 2012, 499, 182-185.	2.2	6
63	<i>ARVCF</i>Genetic Influences on Neurocognitive and Neuroanatomical Intermediate Phenotypes in Chinese Patients With Schizophrenia. Journal of Clinical Psychiatry, 2012, 73, 320-326.	2.2	23
64	An Additional Case of the Recurrent 15q24.1 Microdeletion Syndrome and Review of the Literature. Twin Research and Human Genetics, 2011, 14, 333-339.	0.6	6
65	Methylenetetrahydrofolate reductase (MTHFR) C677T and A1298C polymorphisms and age of onset in schizophrenia: A combined analysis of independent samples. , 2011, 156, 215-224.		19
66	Partial Trisomy 3p and Partial Monosomy 11q Associated with Atrial Septal Defect, Cleft Palate, and Developmental Delay: A Case Report. Cytogenetic and Genome Research, 2011, 134, 319-324.	1.1	1
67	Alcohol and Aldehyde Dehydrogenase Polymorphisms in Chinese and Indian Populations. Substance Use and Misuse, 2010, 45, 1-14.	1.4	18
68	THE METHYLENETETRAHYDROFOLATE REDUCTASE GENE (MTHFR) AND RISK FOR SCHIZOPHRENIA. ARE FUNCTIONAL MTHFR GENE POLYMORPHISMS ASSOCIATED WITH AGE OF ONSET?. Schizophrenia Research, 2010, 117, 336.	2.0	0
69	The influence of ATP-binding cassette sub-family B member -1 (ABCB1) genetic polymorphisms on acute and chronic pain after intrathecal morphine for caesarean section: a prospective cohort study. International Journal of Obstetric Anesthesia, 2010, 19, 254-260.	0.4	54
70	Effect of OPRM variant on labor analgesia and post-cesarean delivery analgesia. International Journal of Obstetric Anesthesia, 2010, 19, 458-459.	0.4	1
71	Ethnicity and OPRM Variant Independently Predict Pain Perception and Patient-Controlled Analgesia Usage for Post-Operative Pain. Molecular Pain, 2009, 5, 1744-8069-5-32.	2.1	107
72	Ethnic Differences in Pain Perception and Patient-Controlled Analgesia Usage for Postoperative Pain. Journal of Pain, 2008, 9, 849-855.	1.4	76

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73	Identification of IRF6 gene variants in three families with Van der Woude syndrome. International Journal of Molecular Medicine, 2008, , .	4.0	7
74	A118G Single Nucleotide Polymorphism of Human $\mu$ -Opioid Receptor Gene Influences Pain Perception and Patient-controlled Intravenous Morphine Consumption after Intrathecal Morphine for Postcesarean Analgesia. Anesthesiology, 2008, 109, 520-526.	2.5	267
75	Identification of IRF6 gene variants in three families with Van der Woude syndrome. International Journal of Molecular Medicine, 2008, 21, 747-51.	4.0	11
76	Functional polymorphisms of the cytochrome P450 1A2 (CYP1A2) gene and prolonged QTc interval in schizophrenia. Progress in Neuro-Psychopharmacology and Biological Psychiatry, 2007, 31, 1297-1302.	4.8	29
77	Epidemiology of Completed Suicides in Singapore for 2001 and 2002. Crisis, 2007, 28, 148-155.	1.2	13
78	Characterization of frequencies and distribution of single nucleotide insertions/deletions in the human genome. Gene, 2006, 376, 268-280.	2.2	6
79	Singapore Human Mutation/Polymorphism Database: a country-specific database for mutations and polymorphisms in inherited disorders and candidate gene association studies. Human Mutation, 2006, 27, 232-235.	2.5	15
80	Congenital long QT syndromes: clinical features, molecular genetics and genetic testing. Expert Review of Molecular Diagnostics, 2006, 6, 365-374.	3.1	13
81	Combined analysis of 635 patients confirms an age-related association of the serotonin 2A receptor gene with tardive dyskinesia and specificity for the non-orofacial subtype. International Journal of Neuropsychopharmacology, 2005, 8, 411-425.	2.1	109
82	Molecular diagnosis of neurogenetic disorders involving trinucleotide repeat expansions. Expert Review of Molecular Diagnostics, 2005, 5, 101-109.	3.1	8
83	Gender-specific association of insertion/deletion polymorphisms in the nogo gene and chronic schizophrenia. Molecular Brain Research, 2005, 139, 212-216.	2.3	35
84	Identification of human Clock gene variants by denaturing high-performance liquid chromatography. Journal of Human Genetics, 2004, 49, 209-214.	2.3	2
85	Genetic analysis of the thermolabile methylenetetrahydrofolate reductase variant in schizophrenia and mood disorders. Psychiatric Genetics, 2004, 14, 227-231.	1.1	49
86	Polymorphisms of dopamine receptors and tardive dyskinesia among Chinese patients with schizophrenia. American Journal of Medical Genetics Part A, 2003, 116B, 51-54.	2.4	54
87	No evidence of molecular heterosis at the dopamine D2 receptor gene locus for smoking in schizophrenia. American Journal of Medical Genetics Part A, 2003, 120B, 40-41.	2.4	1
88	Mu opioid receptor gene polymorphism and neuroleptic-induced tardive dyskinesia in patients with schizophrenia. Schizophrenia Research, 2003, 65, 61-63.	2.0	15
89	Tumor necrosis factor- $\alpha$ gene promoter polymorphisms in chronic schizophrenia. Biological Psychiatry, 2003, 54, 1205-1211.	1.3	32
90	Mu opioid receptor gene polymorphisms and heroin dependence in Asian populations. NeuroReport, 2003, 14, 569-572.	1.2	167

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91	Case-control and linkage disequilibrium studies of the tryptophan hydroxylase gene polymorphisms and major depressive disorder. <i>Psychiatric Genetics</i> , 2003, 13, 151-154.	1.1	10
92	Smoking and tardive dyskinesia: lack of involvement of the CYP1A2 gene. <i>Journal of Psychiatry and Neuroscience</i> , 2003, 28, 185-9.	2.4	27
93	No evidence for association of the T102C polymorphism in the serotonin type 2A receptor with suicidal behavior in schizophrenia. <i>American Journal of Medical Genetics Part A</i> , 2002, 114, 321-322.	2.4	17
94	Heterozygosities and allelic frequencies of a set of microsatellite markers used for genome-wide scans in a Chinese population. <i>Journal of Human Genetics</i> , 2002, 47, 0623-0631.	2.3	3
95	Susceptibility to neuroleptic-induced tardive dyskinesia and the T102C polymorphism in the serotonin type 2A receptor. <i>Biological Psychiatry</i> , 2001, 50, 144-147.	1.3	84
96	Tardive dyskinesia is not associated with the serotonin gene polymorphism (5-HTTLPR) in Chinese. <i>American Journal of Medical Genetics Part A</i> , 2000, 96, 712-715.	2.4	25
97	On the Monophyly of the Agamid Genus <i>Gonocephalus</i> Kaup, 1825 (Reptilia: Squamata). A Chromosomal Perspective.. <i>Current Herpetology</i> , 2000, 19, 71-79.	0.5	5
98	Attempted suicide and polymorphism of the serotonin transporter gene in Chinese patients with schizophrenia. <i>Psychiatry Research</i> , 2000, 97, 101-106.	3.3	49
99	Asthma and TNF variants in Chinese and Malays. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 1999, 54, 402-402.	5.7	14
100	IL-4R $\beta$ gene Ile50Val polymorphism. <i>Allergy: European Journal of Allergy and Clinical Immunology</i> , 1999, 54, 1005-1007.	5.7	2
101	Interleukin-4 receptor variant Q576R: ethnic differences and association with atopy. <i>Clinical Genetics</i> , 1999, 56, 333-334.	2.0	5
102	Evidence for an association between heroin dependence and a VNTR polymorphism at the serotonin transporter locus. <i>Molecular Psychiatry</i> , 1999, 4, 215-217.	7.9	31
103	Novel brain-specific bovine cDNA for a developmentally regulated mRNA encoding a putative new member of the leucine-rich glycoprotein (LRG) family. <i>Neurochemical Research</i> , 1992, 17, 907-916.	3.3	6
104	Stones in Horseshoe Kidneys: Results of Treatment by Extracorporeal Shock Wave Lithotripsy and Endourology. <i>Journal of Urology</i> , 1991, 146, 1213-1215.	0.4	49
105	Novel Variants and Clinical Characteristics of 16 Patients from Southeast Asia with Genetic Variants in Neurofibromin-1. <i>Journal of Pediatric Genetics</i> , 0, , .	0.7	0
106	Fibrous dysplasia in cardiofaciocutaneous syndrome: A case report and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 0, , .	1.2	1