Brian Hon Yin Chung

List of Publications by Year
in descending order

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124 papers	4,661 citations	159585 30 h-index	128289 60 g-index
130 all docs	130 docs citations	130 times ranked	8842 citing authors

#	Article	IF	Citations
1	Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 2020, 180, 568-584.e23.	28.9	1,422
2	Association ofMTORMutations With Developmental Brain Disorders, Including Megalencephaly, Focal Cortical Dysplasia, and Pigmentary Mosaicism. JAMA Neurology, 2016, 73, 836.	9.0	234
3	Practical guidelines for managing adults with 22q11.2 deletion syndrome. Genetics in Medicine, 2015, 17, 599-609.	2.4	222
4	Spinal Muscular Atrophy: Survival Pattern and Functional Status. Pediatrics, 2004, 114, e548-e553.	2.1	142
5	A Modified Screening Tool for Autism (Checklist for Autism in Toddlers [CHAT-23]) for Chinese Children. Pediatrics, 2004, 114, e166-e176.	2.1	132
6	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
7	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	6.2	88
8	Relationship between five common viruses and febrile seizure in children. Archives of Disease in Childhood, 2007, 92, 589-593.	1.9	85
9	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
10	Identification of mutations in the PI3K-AKT-mTOR signalling pathway in patients with macrocephaly and developmental delay and/or autism. Molecular Autism, 2017, 8, 66.	4.9	85
11	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
12	Defects in the Cell Signaling Mediator Î²-Catenin Cause the Retinal Vascular Condition FEVR. American Journal of Human Genetics, 2017, 100, 960-968.	6.2	74
13	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
14	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
15	Clinical reappraisal of <scp>SHORT</scp> syndrome with <scp>PIK3R1</scp> mutations: toward recommendation for molecular testing and management. Clinical Genetics, 2016, 89, 501-506.	2.0	66
16	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
17	A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis. Npj Genomic Medicine, 2020, 5, 37.	3.8	54
18	From VACTERLâ€H to heterotaxy: Variable expressivity of ZIC3â€”related disorders. American Journal of Medical Genetics, Part A, 2011, 155, 1123-1128.	1.2	52

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19	Clinical delineation of the PACS1â€related syndromeâ€"Report on 19 patients. American Journal of Medical Genetics, Part A, 2016, 170, 670-675.	1.2	44
20	Prevalence of Neuromuscular Diseases in Chinese Children: A Study in Southern China. Journal of Child Neurology, 2003, 18, 217-219.	1.4	42
21	Febrile Seizures in Southern Chinese Children: Incidence and Recurrence. Pediatric Neurology, 2006, 34, 121-126.	2.1	42
22	Impact of COVID-19 pandemic on patients with rare disease in Hong Kong. European Journal of Medical Genetics, 2020, 63, 104062.	1.3	42
23	Variable brain phenotype primarily affects the brainstem and cerebellum in patients with osteogenesis imperfecta caused by recessiveWNT1mutations. Journal of Medical Genetics, 2016, 53, 427-430.	3.2	41
24	Whole-Genome Array CGH Evaluation for Replacing Prenatal Karyotyping in Hong Kong. PLoS ONE, 2014, 9, e87988.	2.5	41
25	Genome-Wide DNA Methylation Analysis of Chinese Patients with Systemic Lupus Erythematosus Identified Hypomethylation in Genes Related to the Type I Interferon Pathway. PLoS ONE, 2017, 12, e0169553.	2.5	40
26	Rapid whole-exome sequencing facilitates precision medicine in paediatric rare disease patients and reduces healthcare costs. The Lancet Regional Health - Western Pacific, 2020, 1, 100001.	2.9	40
27	Healthcare burden of rare diseases in Hong Kong â€“ adopting ORPHAcodes in ICD-10 based healthcare administrative datasets. Orphanet Journal of Rare Diseases, 2018, 13, 147.	2.7	39
28	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
29	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
30	Somatic PIK3CA mutations in seven patients with PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2017, 173, 978-984.	1.2	37
31	Copy number variation and autism: New insights and clinical implications. Journal of the Formosan Medical Association, 2014, 113, 400-408.	1.7	36
32	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
33	Prenatal Tobacco Exposure Shortens Telomere Length in Children. Nicotine and Tobacco Research, 2017, 19, 111-118.	2.6	32
34	Identifying the genetic causes for prenatally diagnosed structural congenital anomalies (SCAs) by whole-exome sequencing (WES). BMC Medical Genomics, 2018, 11, 93.	1.5	32
35	Panel-Based Exome Sequencing for Neuromuscular Disorders as a Diagnostic Service. Journal of Neuromuscular Diseases, 2019, 6, 241-258.	2.6	32
36	Mutations in Spliceosomal Genes PPIL1 and PRP17 Cause Neurodegenerative Pontocerebellar Hypoplasia with Microcephaly. Neuron, 2021, 109, 241-256.e9.	8.1	31

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37	Okurâ€€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. Clinical Genetics, 2018, 93, 880-890.	2.0	30
38	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
39	Experience of chromosomal microarray applied in prenatal and postnatal settings in Hong Kong. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 196-207.	1.6	29
40	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
41	Cell lineage-specific genome-wide DNA methylation analysis of patients with paediatric-onset systemic lupus erythematosus. Epigenetics, 2019, 14, 341-351.	2.7	28
42	Rare SUZ12 variants commonly cause an overgrowth phenotype. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 532-547.	1.6	23
43	A thematic study: impact of COVID-19 pandemic on rare disease organisations and patients across ten jurisdictions in the Asia Pacific region. Orphanet Journal of Rare Diseases, 2021, 16, 119.	2.7	22
44	Exome sequencing identifies molecular diagnosis in children with drugâ€resistant epilepsy. Epilepsia Open, 2019, 4, 63-72.	2.4	21
45	Causal somatic mutations in urine DNA from persons with the CLOVES subgroup of the PIK3CAâ€related overgrowth spectrum. Clinical Genetics, 2018, 93, 1075-1080.	2.0	20
46	The Clinical Impact of Chromosomal Microarray on Paediatric Care in Hong Kong. PLoS ONE, 2014, 9, e109629.	2.5	20
47	Identification of novel FBN1 and TGFBR2 mutations in 65 probands with Marfan syndrome or Marfanâ€like phenotypes. American Journal of Medical Genetics, Part A, 2009, 149A, 1452-1459.	1.2	19
48	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
49	Limb body wall complex, amniotic band sequence, or new syndrome caused by mutation in IQ Motif containing K (IQCK)?. Molecular Genetics & Genomic Medicine, 2015, 3, 424-432.	1.2	17
50	Integrating Functional Analysis in the Next-Generation Sequencing Diagnostic Pipeline of RASopathies. Scientific Reports, 2018, 8, 2421.	3.3	17
51	Actionable pharmacogenetic variants in Hong Kong Chinese exome sequencing data and projected prescription impact in the Hong Kong population. PLoS Genetics, 2021, 17, e1009323.	3.5	17
52	Mechanisms for the Generation of Two Quadruplications Associated with Split-Hand Malformation. Human Mutation, 2016, 37, 160-164.	2.5	16
53	Use of clinical chromosomal microarray in Chinese patients with autism spectrum disorderâ€”implications of a copy number variation involving DPP10. Molecular Autism, 2017, 8, 31.	4.9	16
54	A recurrent de novo DYNC1H1 tail domain mutation causes spinal muscular atrophy with lower extremity predominance, learning difficulties and mild brain abnormality. Neuromuscular Disorders, 2018, 28, 750-756.	0.6	16

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55	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	1.2	16
56	A report of three families with FBN1- related acromelic dysplasias and review of literature for genotype-phenotype correlation in geleophysic dysplasia. European Journal of Medical Genetics, 2018, 61, 219-224.	1.3	15
57	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
58	Diagnostic value of wholeâ€exome sequencing in Chinese pediatricâ€onset neuromuscular patients. Molecular Genetics & Genomic Medicine, 2020, 8, e1205.	1.2	15
59	Prenatal and postnatal diagnosis of <scp>Schuursâ€Hoeijmakers</scp> syndrome: Case series and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 384-389.	1.2	15
60	Exome sequencing in paediatric patients with movement disorders. Orphanet Journal of Rare Diseases, 2021, 16, 32.	2.7	15
61	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
62	<scp>CFTR</scp> founder mutation causes protein trafficking defects in Chinese patients with cystic fibrosis. Molecular Genetics & Genomic Medicine, 2017, 5, 40-49.	1.2	14
63	Phenotypic and mutational spectrum of 21 Chinese patients with AlstrÃ¶m syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 279-288.	1.2	14
64	Rare versus common diseases: a false dichotomy in precision medicine. Npj Genomic Medicine, 2021, 6, 19.	3.8	14
65	The epileptology of GNB5 encephalopathy. Epilepsia, 2019, 60, e121-e127.	5.1	13
66	Mowatâ€“Wilson syndrome in a Chinese population: A case series. American Journal of Medical Genetics, Part A, 2020, 182, 1336-1341.	1.2	13
67	Agenesis of the corpus callosum, developmental delay, autism spectrum disorder, facial dysmorphism, and posterior polymorphous corneal dystrophy associated with ZEB1 gene deletion. American Journal of Medical Genetics, Part A, 2017, 173, 2467-2471.	1.2	13
68	Validation and application of health utilities index in Chinese subjects with down syndrome. Health and Quality of Life Outcomes, 2014, 12, 144.	2.4	12
69	The KLHL40 c.1516A>C is a Chineseâ€specific founder mutation causing nemaline myopathy 8: Report of six patients with preâ€and postnatal phenotypes. Molecular Genetics & Genomic Medicine, 2020, 8, e1229.	1.2	12
70	Exome sequencing for paediatric-onset diseases: impact of the extensive involvement of medical geneticists in the diagnostic odyssey. Npj Genomic Medicine, 2018, 3, 19.	3.8	11
71	A significant inflation in TGM6 genetic risk casts doubt in its causation in spinocerebellar ataxia type 35. Parkinsonism and Related Disorders, 2019, 63, 42-45.	2.2	11
72	Delineation of molecular findings by whole-exome sequencing for suspected cases of paediatric-onset mitochondrial diseases in the Southern Chinese population. Human Genomics, 2020, 14, 28.	2.9	11

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73	Inherited thrombophilic factors do not increase central venous catheter blockage in children with malignancy. Pediatric Blood and Cancer, 2008, 51, 509-512.	1.5	10
74	Hospital mortality in patients with rare diseases during pandemics: lessons learnt from the COVID-19 and SARS pandemics. Orphanet Journal of Rare Diseases, 2021, 16, 361.	2.7	10
75	<scp>CTNNB1</scp>â€related neurodevelopmental disorder in a Chinese population: A case series. American Journal of Medical Genetics, Part A, 2022, 188, 130-137.	1.2	10
76	Biallelic <scp>TMEM260</scp> variants cause truncus arteriosus, with or without renal defects. Clinical Genetics, 2022, 101, 127-133.	2.0	10
77	Expanded Praderâ€“Willi syndrome due to chromosome 15q11.2â€“14 deletion: Report and a review of literature. American Journal of Medical Genetics, Part A, 2013, 161, 1309-1318.	1.2	9
78	Training in clinical genetics and genetic counseling in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 177-186.	1.6	9
79	Perception of personalized medicine, pharmacogenomics, and genetic testing among undergraduates in Hong Kong. Human Genomics, 2021, 15, 54.	2.9	9
80	De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033.	3.8	8
81	Megaconial congenital muscular dystrophy: Same novel homozygous mutation in CHKB gene in two unrelated Chinese patients. Neuromuscular Disorders, 2020, 30, 47-53.	0.6	8
82	Client Service Receipt Inventory as a standardised tool for measurement of socio-economic costs in the rare genetic disease population (CSRI-Ra). Scientific Reports, 2021, 11, 23837.	3.3	8
83	Overgrowth with increased proliferation of fibroblast and matrix metalloproteinase activity related to reduced TIMP1: A newly recognized syndrome?. American Journal of Medical Genetics, Part A, 2012, 158A, 2373-2381.	1.2	7
84	A second report of p.Pro986Leu variant in COL2A1â€phenotypic overlap with SEDC and other forms of type II collagenopathies. American Journal of Medical Genetics, Part A, 2013, 161, 918-920.	1.2	7
85	A novel patient with an attenuated Costello syndrome phenotype due to an HRAS mutation affecting codon 146â€”Literature review and update. American Journal of Medical Genetics, Part A, 2017, 173, 1109-1114.	1.2	7
86	Mosaic KRAS mutation in a patient with encephalocraniocutaneous lipomatosis and renovascular hypertension. American Journal of Medical Genetics, Part A, 2018, 176, 2523-2527.	1.2	7
87	Genetic landscape of RASopathies in Chinese: Three decades' experience in Hong Kong. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 208-217.	1.6	7
88	Myhre syndrome: a report of six Chinese patients and literature review. Clinical Dysmorphology, 2019, 28, 143-148.	0.3	7
89	Heterozygous <scp>NOTCH1</scp> deletion associated with variable congenital heart defects. Clinical Genetics, 2021, 99, 836-841.	2.0	7
90	Invasive cerebral phaeohyphomycosis in a Chinese boy with CARD9 deficiency and showing unique radiological features, managed with surgical excision and antifungal treatment. International Journal of Infectious Diseases, 2021, 107, 59-61.	3.3	7

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91	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
92	Paternal uniparental disomy of chromosome 19 in a pair of monochorionic diamniotic twins with dysmorphic features and developmental delay. Journal of Medical Genetics, 2018, 55, 847-852.	3.2	6
93	Comprehensive analysis of recessive carrier status using exome and genome sequencing data in 1543 Southern Chinese. Npj Genomic Medicine, 2022, 7, 23.	3.8	6
94	Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Human Mutation, 2022, 43, 582-594.	2.5	6
95	Hand and fibrillinâ€1 deposition abnormalities in Loeysâ€“Dietz syndromeâ€”expanding the clinical spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 461-466.	1.2	5
96	Further expanding the clinical phenotype in Bainbridge-Ropers syndrome and dissecting genotype-phenotype correlation in the ASXL3 mutational cluster regions. European Journal of Medical Genetics, 2021, 64, 104107.	1.3	5
97	Coffinâ€“Lowry syndrome in Chinese. American Journal of Medical Genetics, Part A, 2019, 179, 2043-2048.	1.2	4
98	Monoallelic Mutations in CC2D1A Suggest a Novel Role in Human Heterotaxy and Ciliary Dysfunction. Circulation Genomic and Precision Medicine, 2020, 13, e003000.	3.6	4
99	A case of G1013R FBN1 mutation: A potential genotypeâ€“phenotype correlation in severe Marfan syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1329-1335.	1.2	4
100	Evaluating the Clinical Utility of Genome Sequencing for Cytogenetically Balanced Chromosomal Abnormalities in Prenatal Diagnosis. Frontiers in Genetics, 2020, 11, 620162.	2.3	4
101	Preparing genomic revolution: Attitudes, clinical practice, and training needs in delivering genetic counseling in primary care in Hong Kong and Shenzhen, China. Molecular Genetics & Genomic Medicine, 2021, 9, e1702.	1.2	4
102	Recurrent de novo missense variants in GNB2 can cause syndromic intellectual disability. Journal of Medical Genetics, 2022, 59, 511-516.	3.2	4
103	Oâ€™Donnell-Luria-Rodan syndrome: description of a second multinational cohort and refinement of the phenotypic spectrum. Journal of Medical Genetics, 2021, , jmedgenet-2020-107470.	3.2	4
104	Evaluating the Health-Related Quality of Life of the Rare Disease Population in Hong Kong Using EQ-5D 3-Level. Value in Health, 2022, 25, 1624-1633.	0.3	4
105	Spread of X inactivation on chromosome 15 is associated with a more severe phenotype in a girl with an unbalanced t(X; 15) translocation. American Journal of Medical Genetics, Part A, 2014, 164, 2521-2528.	1.2	3
106	Headache in a Child with Pseudohypoparathyroidism: An Alarming Symptom Not to Miss. Case Reports in Endocrinology, 2020, 2020, 1-4.	0.4	3
107	Actionable secondary findings in 1116 Hong Kong Chinese based on exome sequencing data. Journal of Human Genetics, 2021, 66, 637-641.	2.3	3
108	Reply: MN1 gene loss-of-function mutation causes cleft palate in a pedigree. Brain, 2021, 144, e19-e19.	7.6	3

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109	Understanding and perception of directâ€toâ€consumer genetic testing in Hong Kong. Journal of Genetic Counseling, 2021, 30, 1640-1648.	1.6	3
110	Elicitation of childrenâ€™s understanding of information in pediatric genetic counseling encounters: A discourseâ€oriented perspective. Journal of Genetic Counseling, 2021, , .	1.6	2
111	Alportâ€™s syndrome: case of a giant esophageal tumor. Esophagus, 2013, 10, 114-117.	1.9	1
112	Prenatal diagnosis of agenesis of the corpus callosum and cerebellar vermian hypoplasia associated with a microdeletion on chromosome 1p32a. Case Reports in Perinatal Medicine, 2013, 2, .	0.1	1
113	Development of clinical genetics in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 150-154.	1.6	1
114	Whole exome sequencing for prenatal diagnosis of CHARGE syndrome: a case report. , 2019, 19, 125-128.		1
115	MG-106â€…Global developmental delay and characteristic facial features associated with pacs1 gene mutation â€“ report of two cases. Journal of Medical Genetics, 2015, 52, A1.2-A1.	3.2	0
116	Physical measurements of Chinese children in Hong Kong-A pilot study in preschools and kindergartens. , 2016, 170, 2069-2077.		0
117	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
118	Cover Image, Volume 173A, Number 9, September 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
119	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
120	Increasing prenatal diagnosis of chimeras with the use of noninvasive prenatal screening: Report of two cases. Prenatal Diagnosis, 2021, 41, 697-700.	2.3	0
121	OMIC-07. FEASIBILITY AND UTILITY OF EPIGENOMIC PROFILING FOR CHILDHOOD CNS TUMORS IN HONG KONG. Neuro-Oncology, 2021, 23, i38-i38.	1.2	0
122	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
123	Prenatal presentation in two fetuses with features of Beckwith Wiedemann syndromeâ€”An unexpected diagnosis of androgenetic chimera and its clinical implications. American Journal of Medical Genetics, Part A, 2022, 188, 1562-1567.	1.2	0
124	Infantile to late adulthood onset facioscapulohumeral dystrophy type 1: a case series. , 2021, 27, 444-449.		0