Christopher C Y Mak

List of Publications by Year
in descending order

31 papers	684 citations	623734 14 h-index	610901 24 g-index
31 all docs	31 docs citations	31 times ranked	1442 citing authors

#	Article	IF	Citations
1	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
2	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
3	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
4	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
5	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
6	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
7	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
8	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
9	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. Npj Genomic Medicine, 2019, 4, 18.	3.8	29
10	Cell lineage-specific genome-wide DNA methylation analysis of patients with paediatric-onset systemic lupus erythematosus. Epigenetics, 2019, 14, 341-351.	2.7	28
11	Exome sequencing identifies molecular diagnosis in children with drugâ€resistant epilepsy. Epilepsia Open, 2019, 4, 63-72.	2.4	21
12	Integrating Functional Analysis in the Next-Generation Sequencing Diagnostic Pipeline of RASopathies. Scientific Reports, 2018, 8, 2421.	3.3	17
13	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
14	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
15	Diagnostic value of wholeâ€exome sequencing in Chinese pediatricâ€onset neuromuscular patients. Molecular Genetics & Genomic Medicine, 2020, 8, e1205.	1.2	15
16	Exome sequencing in paediatric patients with movement disorders. Orphanet Journal of Rare Diseases, 2021, 16, 32.	2.7	15
17	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
18	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

#	Article	IF	Citations
19	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
20	Perception of personalized medicine, pharmacogenomics, and genetic testing among undergraduates in Hong Kong. Human Genomics, 2021, 15, 54.	2.9	9
21	De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033.	3.8	8
22	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
23	A case of prenatal isolated talipes and 22q11.2 deletion syndromeâ€"an important chromosomal disorder missed by noninvasive prenatal screening. Prenatal Diagnosis, 2018, 38, 376-378.	2.3	6
24	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
25	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
26	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
27	Evaluating the Clinical Utility of Genome Sequencing for Cytogenetically Balanced Chromosomal Abnormalities in Prenatal Diagnosis. Frontiers in Genetics, 2020, 11, 620162.	2.3	4
28	Actionable secondary findings in 1116 Hong Kong Chinese based on exome sequencing data. Journal of Human Genetics, 2021, 66, 637-641.	2.3	3
29	Reply: MN1 gene loss-of-function mutation causes cleft palate in a pedigree. Brain, 2021, 144, e19-e19.	7.6	3
30	Understanding and perception of directâ€toâ€consumer genetic testing in Hong Kong. Journal of Genetic Counseling, 2021, 30, 1640-1648.	1.6	3
31	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0