

Christopher C Y Mak

List of Publications by Year in descending order

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31
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1442
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#	ARTICLE	IF	CITATIONS
1	Comprehensive analysis of recessive carrier status using exome and genome sequencing data in 1543 Southern Chinese. <i>Npj Genomic Medicine</i> , 2022, 7, 23.	3.8	6
2	Actionable secondary findings in 1116 Hong Kong Chinese based on exome sequencing data. <i>Journal of Human Genetics</i> , 2021, 66, 637-641.	2.3	3
3	Reply: <i>MN1</i> gene loss-of-function mutation causes cleft palate in a pedigree. <i>Brain</i> , 2021, 144, e19-e19.	7.6	3
4	Exome sequencing in paediatric patients with movement disorders. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 32.	2.7	15
5	Actionable pharmacogenetic variants in Hong Kong Chinese exome sequencing data and projected prescription impact in the Hong Kong population. <i>PLoS Genetics</i> , 2021, 17, e1009323.	3.5	17
6	Understanding and perception of direct-to-consumer genetic testing in Hong Kong. <i>Journal of Genetic Counseling</i> , 2021, 30, 1640-1648.	1.6	3
7	Perception of personalized medicine, pharmacogenomics, and genetic testing among undergraduates in Hong Kong. <i>Human Genomics</i> , 2021, 15, 54.	2.9	9
8	<i>MN1</i> C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. <i>Brain</i> , 2020, 143, 55-68.	7.6	38
9	Phenotypic and mutational spectrum of 21 Chinese patients with <i>Alström</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 279-288.	1.2	14
10	Monoallelic Mutations in <i>CC2D1A</i> Suggest a Novel Role in Human Heterotaxy and Ciliary Dysfunction. <i>Circulation Genomic and Precision Medicine</i> , 2020, 13, e003000.	3.6	4
11	A three-year follow-up study evaluating clinical utility of exome sequencing and diagnostic potential of reanalysis. <i>Npj Genomic Medicine</i> , 2020, 5, 37.	3.8	54
12	Delineation of molecular findings by whole-exome sequencing for suspected cases of paediatric-onset mitochondrial diseases in the Southern Chinese population. <i>Human Genomics</i> , 2020, 14, 28.	2.9	11
13	Rapid whole-exome sequencing facilitates precision medicine in paediatric rare disease patients and reduces healthcare costs. <i>The Lancet Regional Health - Western Pacific</i> , 2020, 1, 100001.	2.9	40
14	Diagnostic value of whole-exome sequencing in Chinese pediatric-onset neuromuscular patients. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1205.	1.2	15
15	Evaluating the Clinical Utility of Genome Sequencing for Cytogenetically Balanced Chromosomal Abnormalities in Prenatal Diagnosis. <i>Frontiers in Genetics</i> , 2020, 11, 620162.	2.3	4
16	Primary coenzyme Q10 deficiency-7: expanded phenotypic spectrum and a founder mutation in southern Chinese. <i>Npj Genomic Medicine</i> , 2019, 4, 18.	3.8	29
17	Genetic landscape of RASopathies in Chinese: Three decades' experience in Hong Kong. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019, 181, 208-217.	1.6	7
18	Cell lineage-specific genome-wide DNA methylation analysis of patients with paediatric-onset systemic lupus erythematosus. <i>Epigenetics</i> , 2019, 14, 341-351.	2.7	28

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19	SLC35A2â€œCDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
20	Exome sequencing identifies molecular diagnosis in children with drugâ€œresistant epilepsy. Epilepsia Open, 2019, 4, 63-72.	2.4	21
21	A case of prenatal isolated talipes and 22q11.2 deletion syndromeâ€œan important chromosomal disorder missed by noninvasive prenatal screening. Prenatal Diagnosis, 2018, 38, 376-378.	2.3	6
22	Integrating Functional Analysis in the Next-Generation Sequencing Diagnostic Pipeline of RASopathies. Scientific Reports, 2018, 8, 2421.	3.3	17
23	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
24	Paternal uniparental disomy of chromosome 19 in a pair of monozygotic diamniotic twins with dysmorphic features and developmental delay. Journal of Medical Genetics, 2018, 55, 847-852.	3.2	6
25	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
26	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
27	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
28	Use of clinical chromosomal microarray in Chinese patients with autism spectrum disorderâ€œimplications of a copy number variation involving DPP10. Molecular Autism, 2017, 8, 31.	4.9	16
29	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
30	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
31	De novo large rare copy-number variations contribute to conotruncal heart disease in Chinese patients. Npj Genomic Medicine, 2016, 1, 16033.	3.8	8