Luk Ho-Ming

List of Publications by Year
in descending order

24 papers	283 citations	1307594 7 h-index	940533 16 g-index
24 all docs	24 docs citations	24 times ranked	684 citing authors

#	Article	IF	Citations
1	Congenital Titinopathy: Comprehensive characterization and pathogenic insights. Annals of Neurology, 2018, 83, 1105-1124.	5.3	93
2	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
3	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
4	Rubinsteinâ€“Taybi syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 2939-2950.	1.2	16
5	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
6	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
7	Clinical and molecular characterization of Beckwith-Wiedemann syndrome in a Chinese population. Journal of Pediatric Endocrinology and Metabolism, 2017, 30, 89-95.	0.9	10
8	Adult Chinese twins with Kennyâ€“Caffey syndrome type 2: A potential ageâ€dependent phenotype and review of literature. American Journal of Medical Genetics, Part A, 2020, 185, 636-646.	1.2	9
9	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
10	Evolving clinical manifestations of mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome: From infancy to adulthood in a 31â€yearâ€old woman. American Journal of Medical Genetics, Part A, 2021, 185, 995-998.	1.2	7
11	CHARGE syndrome in nine patients from China. American Journal of Medical Genetics, Part A, 2020, 182, 15-19.	1.2	6
12	Clinical and molecular characterization study of Chinese Kabuki syndrome in Hong Kong. American Journal of Medical Genetics, Part A, 2021, 185, 675-686.	1.2	6
13	<scp>Rubinsteinâ€Taybi</scp> syndrome in Chinese population with four novel mutations. American Journal of Medical Genetics, Part A, 2021, 185, 267-273.	1.2	5
14	Genotype and phenotype in 18 Chinese patients with Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2250-2261.	1.2	5
15	Coffinâ€“Lowry syndrome in Chinese. American Journal of Medical Genetics, Part A, 2019, 179, 2043-2048.	1.2	4
16	Praderâ€“Willi Syndrome: 16-Year Experience in Hong Kong. Journal of Genetics and Genomics, 2012, 39, 191-194.	3.9	3
17	Adult Prader-Willi Syndrome: An Update on Management. Case Reports in Genetics, 2016, 2016, 1-3.	0.2	3
18	An adult Chinese patient with developmental delay with short stature, dysmorphic features, and sparse hair (<scp>Louksâ€Innes</scp> syndrome). American Journal of Medical Genetics, Part A, 2021, 185, 1925-1931.	1.2	3

#	Article	IF	Citations
19	<scp>KBG</scp> syndrome in a Chinese population: A case series. American Journal of Medical Genetics, Part A, 2022, , .	1.2	3
20	Extending the phenotype of DeSantoâ€Shinawi syndrome: A case report and literature review. American Journal of Medical Genetics, Part A, 2021, , .	1.2	1
21	The first case report of StrÃ¸mme syndrome in a Chinese patient: Expanding the phenotype and literature review. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
22	Successful Treatment of Drug-Resistant Seizures Secondary to Ring 20 Mosaicism with Perampanel as an Add-On Antiepileptic Drug. Case Reports in Pediatrics, 2022, 2022, 1-6.	0.4	1
23	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
24	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