

# Chloe Mak

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/7860442/publications.pdf>

Version: 2024-02-01

10  
papers

216  
citations

1478505

6  
h-index

1474206

9  
g-index

11  
all docs

11  
docs citations

11  
times ranked

392  
citing authors

#	ARTICLE	IF	CITATIONS
1	Inborn errors of metabolism and expanded newborn screening: review and update. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2013, 50, 142-162.	6.1	142
2	Young-Onset Parkinsonism in a Hong Kong Chinese Man With Adult-Onset Hallervordenâ€“Spatz Syndrome. <i>International Journal of Neuroscience</i> , 2011, 121, 224-227.	1.6	20
3	Biochemical and molecular diagnosis of tyrosinemia type I with two novel FAH mutations in a Hong Kong chinese patient: Recommendation for expanded newborn screening in Hong Kong. <i>Clinical Biochemistry</i> , 2013, 46, 155-159.	1.9	15
4	First case of genetically confirmed CLN3 disease in Chinese with cDNA sequencing revealing pathogenicity of a novel splice site variant. <i>Clinica Chimica Acta</i> , 2018, 486, 151-155.	1.1	12
5	Genetic diagnosis of CADASIL in three Hong Kong Chinese patients: A novel mutation within the intracellular domain of NOTCH3. <i>Journal of Clinical Neuroscience</i> , 2018, 56, 95-100.	1.5	9
6	Personalized Medicine Switching from Insulin to Sulfonylurea in Permanent Neonatal Diabetes Mellitus Dictated by a Novel Activating ABCC8 Mutation. <i>Diagnostic Molecular Pathology</i> , 2012, 21, 56-59.	2.1	6
7	Correlation study between spot urine protein-to-creatinine ratio and 24-hour urine protein measurement in 174 patients for proteinuria assessment. <i>Hong Kong Journal of Nephrology</i> , 2011, 13, 51-54.	0.0	5
8	Bi-variate approach to negative interference of bilirubin towards an acetaminophen assay. <i>Clinical Biochemistry</i> , 2015, 48, 186-188.	1.9	5
9	A rare variant of transthyretin-related amyloidosis associated with exclusive cardiomyopathy in a Hong Kong Chinese patient. <i>Journal of Cardiology Cases</i> , 2018, 18, 185-188.	0.5	2
10	Electronic chemical pathology consultation service and dried blood spot metabolic screening in hospital patients. <i>Journal of Clinical Pathology</i> , 2012, 65, 1141-1145.	2.0	0