Chloe Mak

List of Publications by Year in descending order

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1478505 1474206 10 216 6 9 citations h-index g-index papers 11 11 11 392 citing authors docs citations times ranked all docs

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