Lock Hock Ngu

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3542712/publications.pdf

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16	201	933447 10	1058476 14
papers	citations	h-index	g-index

18	18	18	426
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Labruneâ€™s Syndrome Presenting With Stereotypy-Like Movements and Psychosis: A Case Report and Review. Journal of Movement Disorders, 2022, 15, 162-166.	1.3	5
2	A comparison of self-evaluated survey and work sampling approach for estimating patient-care unit cost multiplier in genetic nursing activities. Asian Nursing Research, 2022, , .	1.4	0
3	Efficient detection of copyâ€number variations using exome data: Batchâ€and sexâ€based analyses. Human Mutation, 2021, 42, 50-65.	2.5	18
4	Identification of mutations in Malaysian patients with argininosuccinate lyase (ASL) deficiency. Molecular Genetics and Metabolism Reports, 2019, 21, 100525.	1.1	1
5	Clinical, biochemical and genetic profiles of patients with mucopolysaccharidosis type IVA (Morquio A) Tj ETQq1 1 0.784314 rgBT /Over Diseases, 2019, 14, 143.	2.7	9
6	Fructose-1,6-bisphosphatase deficiency as aÂcause of recurrent hypoglycemia and metabolic acidosis: Clinical and molecular findings in Malaysian patients. Pediatrics and Neonatology, 2018, 59, 397-403.	0.9	13
7	Mutation Study of Malaysian Patients with Ornithine Transcarbamylase Deficiency: Clinical, Molecular, and Bioinformatics Analyses of Two Novel Missense Mutations of the OTC Gene. BioMed Research International, 2018, 2018, 1-15.	1.9	2
8	Combination of Multiple Ligation-Dependent Probe Amplification and Illumina MiSeq Amplicon Sequencing for TSC1/TSC2 Gene Analyses in Patients with Tuberous Sclerosis Complex. Journal of Molecular Diagnostics, 2017, 19, 265-276.	2.8	10
9	Mutations in mitochondrial complex I assembly factor NDUFAF3 cause Leigh syndrome. Molecular Genetics and Metabolism, 2017, 120, 243-246.	1.1	21
10	Pilot study of newborn screening of inborn error of metabolism using tandem mass spectrometry in Malaysia: outcome and challenges. Journal of Pediatric Endocrinology and Metabolism, 2016, 29, 1031-9.	0.9	33
11	N-Carbamylglutamate Is an Effective Treatment for Acute Neonatal Hyperammonaemia in a Patient with Methylmalonic Aciduria. Neonatology, 2016, 109, 303-307.	2.0	14
12	Two Novel Gross Deletions of TSC2 in Malaysian Patients with Tuberous Sclerosis Complex and TSC2/PKD1 Contiguous Deletion Syndrome. Japanese Journal of Clinical Oncology, 2014, 44, 506-511.	1.3	14
13	Mutational Analyses on X-Linked Adrenoleukodystrophy Reveal a Novel Cryptic Splicing and Three Missense Mutations in the ABCD1 Gene. Pediatric Neurology, 2013, 49, 185-190.	2.1	5
14	A catalytic defect in mitochondrial respiratory chain complex I due to a mutation in NDUFS2 in a patient with Leigh syndrome. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 168-175.	3.8	26
15	Adenylosuccinate lyase deficiency in a Malaysian patient, with novel adenylosuccinate lyase gene mutations. Journal of Inherited Metabolic Disease, 2010, 33, 159-162.	3.6	15
16	Neonatal intrahepatic cholestasis associated with citrin deficiency (NICCD): a case series of 11 Malaysian patients. Journal of Inherited Metabolic Disease, 2010, 33, 489-495.	3.6	15