# Lock Hock Ngu

List of Publications by Year
in descending order



| 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 | 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 |
| 2 | 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 |
| 3 | Mutations in mitochondrial complex I assembly factor NDUFAF3 cause Leigh syndrome. Molecular Genetics and Metabolism, 2017, 120, 243-246. | 1.1 | 21 |
| 4 | Efficient detection of copyâ€•number variations using exome data: Batchâ€•and sexâ€•based analyses. Human Mutation, 2021, 42, 50-65. | 2.5 | 18 |
| 5 | 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 |
| 6 | 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 |
| 7 | 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 |
| 8 | N-Carbamylglutamate Is an Effective Treatment for Acute Neonatal Hyperammonaemia in a Patient with Methylmalonic Aciduria. Neonatology, 2016, 109, 303-307. | 2.0 | 14 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | Identification of mutations in Malaysian patients with argininosuccinate lyase (ASL) deficiency. Molecular Genetics and Metabolism Reports, 2019, 21, 100525. | 1.1 | 1 |
| 16 | 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 |