# Catherine Brunel-Guitton

List of Publications by Year
in descending order



| 28 | 961 | 687220 13 | 526166 27 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 28 | 28 | 28 | 2273 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | LRPPRC and SLIRP Interact in a Ribonucleoprotein Complex That Regulates Posttranscriptional Gene Expression in Mitochondria. Molecular Biology of the Cell, 2010, 21, 1315-1323. | 0.9 | 223 |
| 2 | Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397. | 1.1 | 173 |
| 3 | De Novo Mutations in the Motor Domain of KIF1A Cause Cognitive Impairment, Spastic Paraparesis, Axonal Neuropathy, and Cerebellar Atrophy. Human Mutation, 2015, 36, 69-78. | 1.1 | 114 |
| 4 | The 3â€² addition of CCA to mitochondrial tRNASer(AGY) is specifically impaired in patients with mutations in the tRNA nucleotidyl transferase TRNT1. Human Molecular Genetics, 2015, 24, 2841-2847. | 1.4 | 65 |
| 5 | Mitochondrial Diseases and Cardiomyopathies. Canadian Journal of Cardiology, 2015, 31, 1360-1376. | 0.8 | 55 |
| 6 | Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. Journal of Inherited Metabolic Disease, 2016, 39, 173-188. | 1.7 | 45 |
| 7 | Acute pediatric hyperammonemia: current diagnosis and management strategies. Hepatic Medicine: Evidence and Research, 2018, Volume 10, 105-115. | 0.9 | 45 |
| 8 | Inborn errors of cytoplasmic triglyceride metabolism. Journal of Inherited Metabolic Disease, 2015, 38, 85-98. | 1.7 | 29 |
| 9 | Late-onset nonketotic hyperglycinemia caused by a novel homozygous missense mutation in the GLDC gene. Molecular Genetics and Metabolism, 2011, 103, 193-196. | 0.5 | 24 |
| 10 | Combined malonic and methylmalonic aciduria due to ACSF3 mutations: Benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107-116. | 1.7 | 23 |
| 11 | An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. Human Molecular Genetics, 2015, 24, 4103-4113. | 1.4 | 22 |
| 12 | Normal Cerebrospinal Fluid Pyridoxal 5â€²-Phosphate Level in a PNPO-Deficient Patient with Neonatal-Onset Epileptic Encephalopathy. JIMD Reports, 2015, 22, 67-75. | 0.7 | 21 |
| 13 | Treatment of cobalamin C (cblC) deficiency during pregnancy. Journal of Inherited Metabolic Disease, 2010, 33, 409-412. | 1.7 | 17 |
| 14 | The QuÃ©bec NTBC Study. Advances in Experimental Medicine and Biology, 2017, 959, 187-195. | 0.8 | 15 |
| 15 | Premature Ovarian Failure in French Canadian Leigh Syndrome. Journal of Pediatrics, 2017, 184, 227-229.e1. | 0.9 | 14 |
| 16 | Atypical juvenile presentation of GM2 gangliosidosis AB in a patient compound-heterozygote for c.259G > T and c.164C > T mutations in the GM2A gene. Molecular Genetics and Metabolism Reports, 2017, 11, 24-29. | 0.4 | 12 |
| 17 | Evaluation of the quality of clinical data collection for a pan-Canadian cohort of children affected by inherited metabolic diseases: lessons learned from the Canadian Inherited Metabolic Diseases Research Network. Orphanet Journal of Rare Diseases, 2020, 15, 89. | 1.2 | 11 |
| 18 | A Highly Diverse Portrait: Heterogeneity of Neuropsychological Profiles in cblC Defect. JIMD Reports, 2015, 29, 19-32. | 0.7 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment. JIMD Reports, 2017, 39, 89-96. | 0.7 | 8 |
| 20 | Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex I deficiency. Molecular Genetics and Metabolism Reports, 2019, 18, 32-38. | 0.4 | 8 |
| 21 | Prenatal pleural effusions and chylothorax: An unusual presentation for <scp>CMâ€AVM</scp> syndrome due to <scp>RASA1</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2454-2460. | 0.7 | 7 |
| 22 | Health Care for Mitochondrial Disorders in Canada: A Survey of Physicians. Canadian Journal of Neurological Sciences, 2019, 46, 717-726. | 0.3 | 6 |
| 23 | Enzyme replacement therapy in pediatric patients with Gaucher disease: What should we use as maintenance dosage?. Molecular Genetics and Metabolism, 2009, 96, 73-76. | 0.5 | 5 |
| 24 | Combined malonic and methylmalonic aciduria due to ACSF3 mutations: benign clinical course in an unselected cohort. Journal of Inherited Metabolic Disease, 2019, 42, 107. | 1.7 | 4 |
| 25 | Response to Newman et al.. Genetics in Medicine, 2017, 19, 1380-1380. | 1.1 | 3 |
| 26 | Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). Paediatrics and Child Health, 2019, 24, 270-271. | 0.3 | 2 |
| 27 | Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex 1 deficiency. Molecular Genetics and Metabolism Reports, 2019, 19, 100472. | 0.4 | 1 |
| 28 | Reply. Journal of Pediatrics, 2017, 187, 334-335. | 0.9 | 0 |