Klary E Niezen-Koning

List of Publications by Year in descending order

Source: https://exaly.com/author-pdf/6885682/publications.pdf

Version: 2024-02-01

8 168 6 papers citations h-index

8 8 8 319
all docs docs citations times ranked citing authors

8

g-index

| # | Article | IF | CITATIONS |
|---|--|-----|-----------|
| 1 | Clinical pattern, mutations and in vitro residual activity in 33 patients with severe 5, 10 methylenetetrahydrofolate reductase (MTHFR) deficiency. Journal of Inherited Metabolic Disease, 2016, 39, 115-124. | 3.6 | 52 |
| 2 | Translational Targeted Proteomics Profiling of Mitochondrial Energy Metabolic Pathways in Mouse and Human Samples. Journal of Proteome Research, 2016, 15, 3204-3213. | 3.7 | 40 |
| 3 | Living on the edge: substrate competition explains loss of robustness in mitochondrial fatty-acid oxidation disorders. BMC Biology, 2016, 14, 107. | 3.8 | 27 |
| 4 | Neonatal screening for profound biotinidase deficiency in the Netherlands: consequences and considerations. European Journal of Human Genetics, 2016, 24, 1424-1429. | 2.8 | 17 |
| 5 | Homozygous damaging SOD2 variant causes lethal neonatal dilated cardiomyopathy. Journal of Medical Genetics, 2020, 57, 23-30. | 3.2 | 16 |
| 6 | Dopaminergic and serotonergic alterations in plasma in three groups of dystonia patients. Parkinsonism and Related Disorders, 2021, 91, 48-54. | 2.2 | 8 |
| 7 | The Mitochondrial Epigenome: An Unexplored Avenue to Explain Unexplained Myopathies?. International Journal of Molecular Sciences, 2022, 23, 2197. | 4.1 | 7 |
| 8 | Elevated granulocyte-colony stimulating factor and hematopoietic stem cell mobilization in Niemann-Pick type C1 disease. Journal of Lipid Research, 2022, 63, 100167. | 4.2 | 1 |