# Iria Roca

List of Publications by Year
in descending order



| 12 | 297 | 933447 10 | 1199594 12 |
|---|---|---|---|
| papers | citations | h-index | g-index |

| 12 | 12 | 12 | 605 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Free-access copy-number variant detection tools for targeted next-generation sequencing data. Mutation Research - Reviews in Mutation Research, 2019, 779, 114-125. | 5.5 | 46 |
| 2 | Vitamin and mineral status in patients with hyperphenylalaninemia. Molecular Genetics and Metabolism, 2015, 115, 145-150. | 1.1 | 40 |
| 3 | Rare Variants in 48 Genes Account for 42% of Cases of Epilepsy With or Without Neurodevelopmental Delay in 246 Pediatric Patients. Frontiers in Neuroscience, 2019, 13, 1135. | 2.8 | 39 |
| 4 | Carbohydrate status in patients with phenylketonuria. Orphanet Journal of Rare Diseases, 2018, 13, 103. | 2.7 | 36 |
| 5 | Lipid profile status and other related factors in patients with Hyperphenylalaninaemia. Orphanet Journal of Rare Diseases, 2016, 11, 123. | 2.7 | 26 |
| 6 | A novel missense mutation in GRIN2A causes a nonepileptic neurodevelopmental disorder. Movement Disorders, 2018, 33, 992-999. | 3.9 | 26 |
| 7 | Arterial stiffness assessment in patients with phenylketonuria. Medicine (United States), 2017, 96, e9322. | 1.0 | 19 |
| 8 | Molecular-genetic characterization and rescue of a TSFM mutation causing childhood-onset ataxia and nonobstructive cardiomyopathy. European Journal of Human Genetics, 2017, 25, 153-156. | 2.8 | 17 |
| 9 | Evolution of tyrosinemia type 1 disease in patients treated with nitisinone in Spain. Medicine (United) Tj ETQq1 1 0.784314 rgBT /Ove | 1.0 | 17 |
| 10 | Prioritization of Variants Detected by Next Generation Sequencing According to the Mutation Tolerance and Mutational Architecture of the Corresponding Genes. International Journal of Molecular Sciences, 2018, 19, 1584. | 4.1 | 16 |
| 11 | PattRec: An easy-to-use CNV detection tool optimized for targeted NGS assays with diagnostic purposes. Genomics, 2020, 112, 1245-1256. | 2.9 | 10 |
| 12 | Micronutrient in hyperphenylalaninemia. Data in Brief, 2015, 4, 614-621. | 1.0 | 5 |