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	PattRec: An easy-to-use CNV detection tool optimized for targeted NGS assays with diagnostic purposes. Genomics, 2020, 112, 1245-1256.	2.9	10
2	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
3	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
4	Evolution of tyrosinemia type 1 disease in patients treated with nitisinone in Spain. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.0	17
5	A novel missense mutation in GRIN2A causes a nonepileptic neurodevelopmental disorder. Movement Disorders, 2018, 33, 992-999.	3.9	26
6	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
7	Carbohydrate status in patients with phenylketonuria. Orphanet Journal of Rare Diseases, 2018, 13, 103.	2.7	36
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	Arterial stiffness assessment in patients with phenylketonuria. Medicine (United States), 2017, 96, e9322.	1.0	19
10	Lipid profile status and other related factors in patients with Hyperphenylalaninaemia. Orphanet Journal of Rare Diseases, 2016, 11, 123.	2.7	26
11	Micronutrient in hyperphenylalaninemia. Data in Brief, 2015, 4, 614-621.	1.0	5
12	Vitamin and mineral status in patients with hyperphenylalaninemia. Molecular Genetics and Metabolism, 2015, 115, 145-150.	1.1	40