IÃ±igo Marcos-Alcalde

List of Publications by Year
in descending order

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20 papers	395 citations	933410 10 h-index	794568 19 g-index
20 all docs	20 docs citations	20 times ranked	820 citing authors

#	Article	IF	Citations
1	The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder. Human Mutation, 2022, 43, 266-282.	2.5	7
2	Diversity of mechanisms to control bacterial <scp>GTP</scp> homeostasis by the mutually exclusive binding of adenine and guanine nucleotides to <scp>IMP</scp> dehydrogenase. Protein Science, 2022, 31, e4314.	7.6	9
3	Pathogenic convergence of CNVs in genes functionally associated to a severe neuromotor developmental delay syndrome. Human Genomics, 2021, 15, 11.	2.9	3
4	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
5	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. Scientific Reports, 2021, 11, 15459.	3.3	11
6	Dysfunctional Homozygous VRK1-D263G Variant Impairs the Assembly of Cajal Bodies and DNA Damage Response in Hereditary Spastic Paraplegia. Neurology: Genetics, 2021, 7, e624.	1.9	2
7	Things are not always what they seem: From Cornelia de Lange to KBG phenotype in a girl with genetic variants in NIPBL and ANKRD11. Molecular Genetics & Genomic Medicine, 2021, 9, e1826.	1.2	2
8	MEPSAnd: minimum energy path surface analysis overn-dimensional surfaces. Bioinformatics, 2020, 36, 956-958.	4.1	14
9	VRK1 (Y213H) homozygous mutant impairs Cajal bodies in a hereditary case of distal motor neuropathy. Annals of Clinical and Translational Neurology, 2020, 7, 808-818.	3.7	8
10	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
11	Novel Dominant KCNQ2 Exon 7 Partial In-Frame Duplication in a Complex Epileptic and Neurodevelopmental Delay Syndrome. International Journal of Molecular Sciences, 2020, 21, 4447.	4.1	5
12	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042.	4.1	40
13	A novel RAD21 p.(Gln592del) variant expands the clinical description of Cornelia de Lange syndrome type 4 â€“ Review of the literature. European Journal of Medical Genetics, 2019, 62, 103526.	1.3	12
14	Human Mitochondrial HMG-CoA Synthase Deficiency: Role of Enzyme Dimerization Surface and Characterization of Three New Patients. International Journal of Molecular Sciences, 2018, 19, 1010.	4.1	20
15	Two-step ATP-driven opening of cohesin head. Scientific Reports, 2017, 7, 3266.	3.3	19
16	A nucleotide-controlled conformational switch modulates the activity of eukaryotic IMP dehydrogenases. Scientific Reports, 2017, 7, 2648.	3.3	36
17	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
18	A Practical Quantum Mechanics Molecular Mechanics Method for the Dynamical Study of Reactions in Biomolecules. Advances in Protein Chemistry and Structural Biology, 2015, 100, 67-88.	2.3	5

#	ARTICLE	IF	CITATIONS
19	MEPSA: minimum energy pathway analysis for energy landscapes. Bioinformatics, 2015, 31, 3853-3855.	4.1	84
20	Simulation of Catalytic Water Activation in Mitochondrial F₁-ATPase Using a Hybrid Quantum Mechanics/Molecular Mechanics Approach: An Alternative Role for Î²-Glu 188. Biochemistry, 2013, 52, 959-966.	2.5	6