Juan PiÃ©

List of Publications by Year
in descending order

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69 papers	2,417 citations	257450 24 h-index	223800 46 g-index
71 all docs	71 docs citations	71 times ranked	3228 citing authors

#	Article	IF	Citations
1	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
2	Subclinical myocardial dysfunction is revealed by speckle tracking echocardiography in patients with Cornelia de Lange syndrome. International Journal of Cardiovascular Imaging, 2022, 38, 2291-2302.	0.6	1
3	Schuursâ€“Hoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. Genes, 2021, 12, 738.	2.4	13
4	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
5	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
6	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	2.5	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	High rate of autonomic neuropathy in Cornelia de Lange Syndrome. Orphanet Journal of Rare Diseases, 2021, 16, 458.	2.7	0
9	MEPSAnd: minimum energy path surface analysis overn-dimensional surfaces. Bioinformatics, 2020, 36, 956-958.	4.1	14
10	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	2.0	10
11	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Reports, 2020, 31, 107647.	6.4	36
12	Pathogenic variants in <scp>EP300</scp> and <scp>ANKRD11</scp> in patients with phenotypes overlapping Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1690-1696.	1.2	34
13	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
14	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
15	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
16	The gene encoding the ketogenic enzyme HMGCS2 displays a unique expression during gonad development in mice. PLoS ONE, 2020, 15, e0227411.	2.5	12
17	SÃndrome Cornelia de Lange: tamaÃ±o y funciÃ³n ventricular en seis casos sin cardiopatÃa congÃ©nita. Medicina ClÃnica, 2020, 154, 67-68.	0.6	0
18	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

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19	More Than One HMG-CoA Lyase: The Classical Mitochondrial Enzyme Plus the Peroxisomal and the Cytosolic Ones. International Journal of Molecular Sciences, 2019, 20, 6124.	4.1	14
20	Development, behaviour and autism in individuals with SMC1A variants. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2019, 60, 305-313.	5.2	13
21	Reduced immunoglobulin gene diversity in patients with Cornelia de Lange syndrome. Journal of Allergy and Clinical Immunology, 2018, 141, 408-411.e8.	2.9	6
22	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
23	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
24	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. Human Genetics, 2017, 136, 307-320.	3.8	61
25	Two-step ATP-driven opening of cohesin head. Scientific Reports, 2017, 7, 3266.	3.3	19
26	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
27	SÃndrome de Cornelia de Lange: incidencia de cardiopatÃa congÃ©nita en 149 pacientes. Medicina ClÃnica, 2017, 149, 300-302.	0.6	9
28	mRNA Quantification of NIPBL Isoforms A and B in Adult and Fetal Human Tissues, and a Potentially Pathological Variant Affecting Only Isoform A in Two Patients with Cornelia de Lange Syndrome. International Journal of Molecular Sciences, 2017, 18, 481.	4.1	1
29	Identification and Functional Characterization of Two IntronicNIPBLMutations in Two Patients with Cornelia de Lange Syndrome. BioMed Research International, 2016, 2016, 1-8.	1.9	12
30	Expanding the clinical spectrum of the â€˜<scp>HDAC8</scp>â€™phenotypeâ€™ â€“ implications for molecular diagnostics, counseling and risk prediction. Clinical Genetics, 2016, 89, 564-573.	2.0	38
31	Quantum Mechanics/Molecular Mechanics Free Energy Maps and Nonadiabatic Simulations for a Photochemical Reaction in DNA: Cyclobutane Thymine Dimer. Journal of Physical Chemistry Letters, 2016, 7, 4391-4397.	4.6	20
32	Special cases in Cornelia de Lange syndrome: The Spanish experience. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2016, 172, 198-205.	1.6	19
33	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	2.5	72
34	Effects of detyrosinated tubulin on Na⁺,K⁺â€ATPase activity and erythrocyte function in hypertensive subjects. FEBS Letters, 2015, 589, 364-373.	2.8	16
35	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	2.8	37
36	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

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37	MEPSA: minimum energy pathway analysis for energy landscapes. Bioinformatics, 2015, 31, 3853-3855.	4.1	84
38	Functional Characterization of NIPBL Physiological Splice Variants and Eight Splicing Mutations in Patients with Cornelia de Lange Syndrome. International Journal of Molecular Sciences, 2014, 15, 10350-10364.	4.1	22
39	Somatic mosaicism in a Cornelia de Lange syndrome patient with <scp>NIPBL</scp> mutation identified by different next generation sequencing approaches. Clinical Genetics, 2014, 86, 595-597.	2.0	17
40	Could a patient with <scp>SMC1A</scp> duplication beÂclassified as a human cohesinopathy?. Clinical Genetics, 2014, 85, 446-451.	2.0	12
41	Efficient application of next-generation sequencing for the diagnosis of rare genetic syndromes. Journal of Clinical Pathology, 2014, 67, 1099-1103.	2.0	13
42	<scp>fireball</scp>/<scp>amber</scp>: An Efficient Local-Orbital DFT QM/MM Method for Biomolecular Systems. Journal of Chemical Theory and Computation, 2014, 10, 2185-2193.	5.3	42
43	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	2.9	120
44	Severe ipsilateral musculoskeletal involvement in a Cornelia de Lange patient with a novel NIPBL mutation. European Journal of Medical Genetics, 2014, 57, 503-509.	1.3	8
45	Analysis of aberrant splicing and nonsense-mediated decay of the stop codon mutations c.109G>T and c.504_505delCT in 7 patients with HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2013, 108, 232-240.	1.1	7
46	Carnitine palmitoyltransferase 1C deficiency causes motor impairment and hypoactivity. Behavioural Brain Research, 2013, 256, 291-297.	2.2	38
47	New case of mitochondrial HMG-CoA synthase deficiency. Functional analysis of eight mutations. European Journal of Medical Genetics, 2013, 56, 411-415.	1.3	23
48	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. Journal of Experimental Medicine, 2013, 210, 2503-2513.	8.5	33
49	Characterization of a novel HMG-CoA lyase enzyme with a dual location in endoplasmic reticulum and cytosol. Journal of Lipid Research, 2012, 53, 2046-2056.	4.2	8
50	Involvement of membrane tubulin in erythrocyte deformability and blood pressure. Journal of Hypertension, 2012, 30, 1414-1422.	0.5	23
51	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. BMC Medical Genetics, 2012, 13, 43.	2.1	12
52	The Role of Gln61 in HRas GTP Hydrolysis: A Quantum Mechanics/Molecular Mechanics Study. Biophysical Journal, 2012, 102, 152-157.	0.5	48
53	Characterization of splice variants of the genes encoding human mitochondrial HMG-CoA lyase and HMG-CoA synthase, the main enzymes of the ketogenesis pathway. Molecular Biology Reports, 2012, 39, 4777-4785.	2.3	24
54	Differential HMGâ€CoA lyase expression in human tissues provides clues about 3â€hydroxyâ€3â€methylglutaric aciduria. Journal of Inherited Metabolic Disease, 2010, 33, 405-410.	3.6	20

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55	Mutations and variants in the cohesion factor genes NIPBL, SMC1A, and SMC3 in a cohort of 30 unrelated patients with Cornelia de Lange syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 924-929.	1.2	72
56	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. American Journal of Medical Genetics, Part A, 2010, 152A, 1641-1653.	1.2	75
57	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	2.5	21
58	C-Terminal end and aminoacid Lys48 in HMG-CoA lyase are involved in substrate binding and enzyme activity. Molecular Genetics and Metabolism, 2007, 91, 120-127.	1.1	10
59	Molecular genetics of HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2007, 92, 198-209.	1.1	64
60	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. American Journal of Human Genetics, 2007, 80, 485-494.	6.2	445
61	Skipping of exon 2 and exons 2 plus 3 of HMG-CoA lyase (HL) gene produces the loss of beta sheets 1 and 2 in the recently proposed (beta-alpha)8 TIM Barrel model of HL. Biophysical Chemistry, 2005, 115, 241-245.	2.8	11
62	Structural (Î²Î±)8 TIM Barrel Model of 3-Hydroxy-3-methylglutaryl-Coenzyme A Lyase. Journal of Biological Chemistry, 2003, 278, 29016-29023.	3.4	17
63	The diagnosis of mitochondrial HMG-CoA synthase deficiency. Journal of Pediatrics, 2002, 140, 778-780.	1.8	30
64	Genetic basis of mitochondrial HMG-CoA synthase deficiency. Human Genetics, 2001, 109, 19-23.	3.8	45
65	5-methoxytryptophol preserves hepatic microsomal membrane fluidity during oxidative stress. , 2000, 76, 651-657.		22
66	Role of pinoline and melatonin in stabilizing hepatic microsomal membranes against oxidative stress. Journal of Bioenergetics and Biomembranes, 1999, 31, 609-616.	2.3	92
67	A Nonsense Mutation in the Exon 2 of the 3-Hydroxy- 3-methylglutaryl Coenzyme A Lyase (HL) Gene Producing Three Mature mRNAs Is the Main Cause of 3-Hydroxy-3-methylglutaric Aciduria in European Mediterranean Patients. Archives of Biochemistry and Biophysics, 1998, 349, 129-137.	3.0	26
68	Two Missense Point Mutations in Different Alleles in the 3-Hydroxy-3-methylglutaryl Coenzyme A Lyase Gene Produce 3-Hydroxy-3-methylglutaric Aciduria in a French Patient. Archives of Biochemistry and Biophysics, 1998, 358, 197-203.	3.0	13
69	A nonsense mutation in the 3-hydroxy-3-methylglutaryl-CoA lyase gene produces exon skipping in two patients of different origin with 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. Biochemical Journal, 1997, 323, 329-335.	3.7	39