

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	Mutations in Cohesin Complex Members SMC3 and SMC1A Cause a Mild Variant of Cornelia de Lange Syndrome with Predominant Mental Retardation. <i>American Journal of Human Genetics</i> , 2007, 80, 485-494.	6.2	445
2	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. <i>Nature Reviews Genetics</i> , 2018, 19, 649-666.	16.3	223
3	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. <i>Human Molecular Genetics</i> , 2014, 23, 2888-2900.	2.9	120
4	Role of pinoline and melatonin in stabilizing hepatic microsomal membranes against oxidative stress. <i>Journal of Bioenergetics and Biomembranes</i> , 1999, 31, 609-616.	2.3	92
5	MEPSA: minimum energy pathway analysis for energy landscapes. <i>Bioinformatics</i> , 2015, 31, 3853-3855.	4.1	84
6	Facial diagnosis of mild and variant CdLS: Insights from a dysmorphologist survey. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1641-1653.	1.2	75
7	Mutations and variants in the cohesion factor genes <i>NIPBL</i> , <i>SMC1A</i> , and <i>SMC3</i> in a cohort of 30 unrelated patients with Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 924-929.	1.2	72
8	<i>De Novo</i> Heterozygous Mutations in <i>SMC3</i> Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. <i>Human Mutation</i> , 2015, 36, 454-462.	2.5	72
9	Phenotypes and genotypes in individuals with <i>SMC1A</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2108-2125.	1.2	69
10	Molecular genetics of HMG-CoA lyase deficiency. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 198-209.	1.1	64
11	Mutations in chromatin regulators functionally link Cornelia de Lange syndrome and clinically overlapping phenotypes. <i>Human Genetics</i> , 2017, 136, 307-320.	3.8	61
12	The Role of Gln61 in HRas GTP Hydrolysis: A Quantum Mechanics/Molecular Mechanics Study. <i>Biophysical Journal</i> , 2012, 102, 152-157.	0.5	48
13	Genetic basis of mitochondrial HMG-CoA synthase deficiency. <i>Human Genetics</i> , 2001, 109, 19-23.	3.8	45
14	<i>fireball</i> / <i>amber</i> : An Efficient Local-Orbital DFT QM/MM Method for Biomolecular Systems. <i>Journal of Chemical Theory and Computation</i> , 2014, 10, 2185-2193.	5.3	42
15	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. <i>International Journal of Molecular Sciences</i> , 2020, 21, 1042.	4.1	40
16	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. <i>Biochemical Journal</i> , 1997, 323, 329-335.	3.7	39
17	Carnitine palmitoyltransferase 1C deficiency causes motor impairment and hypoactivity. <i>Behavioural Brain Research</i> , 2013, 256, 291-297.	2.2	38
18	Expanding the clinical spectrum of the <i>HDAC8</i> phenotype™ implications for molecular diagnostics, counseling and risk prediction. <i>Clinical Genetics</i> , 2016, 89, 564-573.	2.0	38

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19	Clinical utility gene card for: Cornelia de Lange syndrome. <i>European Journal of Human Genetics</i> , 2015, 23, 1431-1431.	2.8	37
20	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. <i>Cell Reports</i> , 2020, 31, 107647.	6.4	36
21	Pathogenic variants in <i>EP300</i> and <i>ANKRD11</i> in patients with phenotypes overlapping Cornelia de Lange syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1690-1696.	1.2	34
22	A regulatory role for the cohesin loader NIPBL in nonhomologous end joining during immunoglobulin class switch recombination. <i>Journal of Experimental Medicine</i> , 2013, 210, 2503-2513.	8.5	33
23	The diagnosis of mitochondrial HMG-CoA synthase deficiency. <i>Journal of Pediatrics</i> , 2002, 140, 778-780.	1.8	30
24	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. <i>Archives of Biochemistry and Biophysics</i> , 1998, 349, 129-137.	3.0	26
25	Characterization of splice variants of the genes encoding human mitochondrial HMG-CoA lyase and HMG-CoA synthase, the main enzymes of the ketogenesis pathway. <i>Molecular Biology Reports</i> , 2012, 39, 4777-4785.	2.3	24
26	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. <i>Human Genetics</i> , 2020, 139, 575-592.	3.8	24
27	Involvement of membrane tubulin in erythrocyte deformability and blood pressure. <i>Journal of Hypertension</i> , 2012, 30, 1414-1422.	0.5	23
28	New case of mitochondrial HMG-CoA synthase deficiency. Functional analysis of eight mutations. <i>European Journal of Medical Genetics</i> , 2013, 56, 411-415.	1.3	23
29	5-methoxytryptophol preserves hepatic microsomal membrane fluidity during oxidative stress. , 2000, 76, 651-657.		22
30	Functional Characterization of NIPBL Physiological Splice Variants and Eight Splicing Mutations in Patients with Cornelia de Lange Syndrome. <i>International Journal of Molecular Sciences</i> , 2014, 15, 10350-10364.	4.1	22
31	Ten novel HMGCL mutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. <i>Human Mutation</i> , 2009, 30, E520-E529.	2.5	21
32	Differential HMG-CoA lyase expression in human tissues provides clues about 3-hydroxy-3-methylglutaric aciduria. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 405-410.	3.6	20
33	Quantum Mechanics/Molecular Mechanics Free Energy Maps and Nonadiabatic Simulations for a Photochemical Reaction in DNA: Cyclobutane Thymine Dimer. <i>Journal of Physical Chemistry Letters</i> , 2016, 7, 4391-4397.	4.6	20
34	Human Mitochondrial HMG-CoA Synthase Deficiency: Role of Enzyme Dimerization Surface and Characterization of Three New Patients. <i>International Journal of Molecular Sciences</i> , 2018, 19, 1010.	4.1	20
35	Special cases in Cornelia de Lange syndrome: The Spanish experience. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2016, 172, 198-205.	1.6	19
36	Two-step ATP-driven opening of cohesin head. <i>Scientific Reports</i> , 2017, 7, 3266.	3.3	19

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37	Structural (β ²) ₈ TIM Barrel Model of 3-Hydroxy-3-methylglutaryl-Coenzyme A Lyase. <i>Journal of Biological Chemistry</i> , 2003, 278, 29016-29023.	3.4	17
38	Somatic mosaicism in a Cornelia de Lange syndrome patient with <i>NIPBL</i> mutation identified by different next generation sequencing approaches. <i>Clinical Genetics</i> , 2014, 86, 595-597.	2.0	17
39	Effects of detyrosinated tubulin on Na ⁺ ,K ⁺ -ATPase activity and erythrocyte function in hypertensive subjects. <i>FEBS Letters</i> , 2015, 589, 364-373.	2.8	16
40	MEPSAnd: minimum energy path surface analysis over <i>n</i> -dimensional surfaces. <i>Bioinformatics</i> , 2020, 36, 956-958.	4.1	14
41	More Than One HMG-CoA Lyase: The Classical Mitochondrial Enzyme Plus the Peroxisomal and the Cytosolic Ones. <i>International Journal of Molecular Sciences</i> , 2019, 20, 6124.	4.1	14
42	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. <i>Archives of Biochemistry and Biophysics</i> , 1998, 358, 197-203.	3.0	13
43	Efficient application of next-generation sequencing for the diagnosis of rare genetic syndromes. <i>Journal of Clinical Pathology</i> , 2014, 67, 1099-1103.	2.0	13
44	Development, behaviour and autism in individuals with <i>SMC1A</i> variants. <i>Journal of Child Psychology and Psychiatry and Allied Disciplines</i> , 2019, 60, 305-313.	5.2	13
45	Schuursâ€œHoeijmakers Syndrome (PACS1 Neurodevelopmental Disorder): Seven Novel Patients and a Review. <i>Genes</i> , 2021, 12, 738.	2.4	13
46	Cornelia de Lange syndrome with <i>NIPBL</i> mutation and mosaic Turner syndrome in the same individual. <i>BMC Medical Genetics</i> , 2012, 13, 43.	2.1	12
47	Could a patient with <i>SMC1A</i> duplication be classified as a human cohesinopathy?. <i>Clinical Genetics</i> , 2014, 85, 446-451.	2.0	12
48	Identification and Functional Characterization of Two Intronic <i>NIPBL</i> Mutations in Two Patients with Cornelia de Lange Syndrome. <i>BioMed Research International</i> , 2016, 2016, 1-8.	1.9	12
49	A novel <i>RAD21</i> p.(Gln592del) variant expands the clinical description of Cornelia de Lange syndrome type 4 â€œ Review of the literature. <i>European Journal of Medical Genetics</i> , 2019, 62, 103526.	1.3	12
50	The gene encoding the ketogenic enzyme <i>HMGCS2</i> displays a unique expression during gonad development in mice. <i>PLoS ONE</i> , 2020, 15, e0227411.	2.5	12
51	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. <i>Biophysical Chemistry</i> , 2005, 115, 241-245.	2.8	11
52	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of <i>NIPBL</i> variants in blood. <i>Scientific Reports</i> , 2021, 11, 15459.	3.3	11
53	C-Terminal end and aminoacid Lys48 in HMG-CoA lyase are involved in substrate binding and enzyme activity. <i>Molecular Genetics and Metabolism</i> , 2007, 91, 120-127.	1.1	10
54	Heterozygous de novo variants in <i>CSNK1G1</i> are associated with syndromic developmental delay and autism spectrum disorder. <i>Clinical Genetics</i> , 2020, 98, 571-576.	2.0	10

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55	SÃndrome de Cornelia de Lange: incidencia de cardiopatÃa congÃ©nita en 149 pacientes. Medicina ClÃnica, 2017, 149, 300-302.	0.6	9
56	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
57	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
58	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
59	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
60	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
61	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
62	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
63	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
64	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	2.5	2
65	Things are not always what they seem: From Cornelia de Lange to KBC phenotype in a girl with genetic variants in NIPBL and ANKRD11. Molecular Genetics & Genomic Medicine, 2021, 9, e1826.	1.2	2
66	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
67	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
68	High rate of autonomic neuropathy in Cornelia de Lange Syndrome. Orphanet Journal of Rare Diseases, 2021, 16, 458.	2.7	0
69	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