Beatriz Puisac

List of Publications by Year
in descending order

33	807	471061 17	525886 27
papers	citations	h-index	g-index

33	33	33	1414
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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.	1.4	120
2	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.	0.7	72
3	De NovoHeterozygous Mutations inSMC3Cause a Range of Cornelia de Lange Syndrome-Overlapping Phenotypes. Human Mutation, 2015, 36, 454-462.	1.1	72
4	Molecular genetics of HMG-CoA lyase deficiency. Molecular Genetics and Metabolism, 2007, 92, 198-209.	0.5	64
5	Evaluating Face2Gene as a Tool to Identify Cornelia de Lange Syndrome by Facial Phenotypes. International Journal of Molecular Sciences, 2020, 21, 1042.	1.8	40
6	Clinical utility gene card for: Cornelia de Lange syndrome. European Journal of Human Genetics, 2015, 23, 1431-1431.	1.4	37
7	MAU2 and NIPBL Variants Impair the Heterodimerization of the Cohesin Loader Subunits and Cause Cornelia de Lange Syndrome. Cell Reports, 2020, 31, 107647.	2.9	36
8	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.	0.7	34
9	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.	4.2	33
10	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.	1.0	24
11	New case of mitochondrial HMG-CoA synthase deficiency. Functional analysis of eight mutations. European Journal of Medical Genetics, 2013, 56, 411-415.	0.7	23
12	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.	1.8	22
13	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	1.1	21
14	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.	1.7	20
15	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.	1.8	20
16	Disruption of NIPBL/Scc2 in Cornelia de Lange Syndrome provokes cohesin genome-wide redistribution with an impact in the transcriptome. Nature Communications, 2021, 12, 4551.	5.8	20
17	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.	0.7	19
18	Two-step ATP-driven opening of cohesin head. Scientific Reports, 2017, 7, 3266.	1.6	19

#	Article	IF	Citations
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.	1.8	14
20	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. BMC Medical Genetics, 2012, 13, 43.	2.1	12
21	Identification and Functional Characterization of Two IntronicNIPBLMutations in Two Patients with Cornelia de Lange Syndrome. BioMed Research International, 2016, 2016, 1-8.	0.9	12
22	The gene encoding the ketogenic enzyme HMGCS2 displays a unique expression during gonad development in mice. PLoS ONE, 2020, 15, e0227411.	1.1	12
23	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.	1.5	11
24	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. Scientific Reports, 2021, 11, 15459.	1.6	11
25	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	1.0	10
26	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.	2.0	8
27	Severe ipsilateral musculoskeletal involvement in a Cornelia de Lange patient with a novel NIPBL mutation. European Journal of Medical Genetics, 2014, 57, 503-509.	0.7	8
28	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.	0.5	7
29	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	1.3	2
30	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.	0.6	2
31	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.	1.8	1
32	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.	0.2	1
33	High rate of autonomic neuropathy in Cornelia de Lange Syndrome. Orphanet Journal of Rare Diseases, 2021, 16, 458.	1.2	0