Maria Arnedo

List of Publications by Year
in descending order

17 papers	868 citations	933447 10 h-index	940533 16 g-index
17 all docs	17 docs citations	17 times ranked	1557 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. American Journal of Human Genetics, 2007, 80, 485-494.	6.2	445
2	SAF-A Regulates Interphase Chromosome Structure through Oligomerization with Chromatin-Associated RNAs. Cell, 2017, 169, 1214-1227.e18.	28.9	166
3	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
4	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
5	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
6	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
7	Ten novelHMGCLmutations in 24 patients of different origin with 3-hydroxy-3-methyl-glutaric aciduria. Human Mutation, 2009, 30, E520-E529.	2.5	21
8	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
9	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
10	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. BMC Medical Genetics, 2012, 13, 43.	2.1	12
11	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
12	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
13	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
14	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. Applied Sciences (Switzerland), 2021, 11, 710.	2.5	2
15	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
16	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
17	High rate of autonomic neuropathy in Cornelia de Lange Syndrome. Orphanet Journal of Rare Diseases, 2021, 16, 458.	2.7	0