

Maria Arnedo

List of Publications by Year in descending order

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Version: 2024-02-01

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	Subclinical myocardial dysfunction is revealed by speckle tracking echocardiography in patients with Cornelia de Lange syndrome. <i>International Journal of Cardiovascular Imaging</i> , 2022, 38, 2291-2302.	0.6	1
2	Clinical relevance of postzygotic mosaicism in Cornelia de Lange syndrome and purifying selection of NIPBL variants in blood. <i>Scientific Reports</i> , 2021, 11, 15459.	3.3	11
3	Targeted Gene Sequencing, Bone Health, and Body Composition in Cornelia de Lange Syndrome. <i>Applied Sciences (Switzerland)</i> , 2021, 11, 710.	2.5	2
4	Things are not always what they seem: From Cornelia de Lange to KBG phenotype in a girl with genetic variants in NIPBL and ANKRD11. <i>Molecular Genetics & Genomic Medicine</i> , 2021, 9, e1826.	1.2	2
5	High rate of autonomic neuropathy in Cornelia de Lange Syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 458.	2.7	0
6	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
7	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
8	SAF-A Regulates Interphase Chromosome Structure through Oligomerization with Chromatin-Associated RNAs. <i>Cell</i> , 2017, 169, 1214-1227.e18.	28.9	166
9	Analysis of aberrant splicing and nonsense-mediated decay of the stop codon mutations c.109C>T and c.504_505delCT in 7 patients with HMG-CoA lyase deficiency. <i>Molecular Genetics and Metabolism</i> , 2013, 108, 232-240.	1.1	7
10	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
11	Characterization of a novel HMG-CoA lyase enzyme with a dual location in endoplasmic reticulum and cytosol. <i>Journal of Lipid Research</i> , 2012, 53, 2046-2056.	4.2	8
12	Cornelia de Lange syndrome with NIPBL mutation and mosaic Turner syndrome in the same individual. <i>BMC Medical Genetics</i> , 2012, 13, 43.	2.1	12
13	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
14	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
15	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
16	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
17	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