

Laura AlÃ- as

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

1,021
citations

567281

15
h-index

580821

25
g-index

25
all docs

25
docs citations

25
times ranked

981
citing authors

#	ARTICLE	IF	CITATIONS
1	Correlation between SMA type and SMN2 copy number revisited: An analysis of 625 unrelated Spanish patients and a compilation of 2834 reported cases. <i>Neuromuscular Disorders</i> , 2018, 28, 208-215.	0.6	273
2	Mutation update of spinal muscular atrophy in Spain: molecular characterization of 745 unrelated patients and identification of four novel mutations in the SMN1 gene. <i>Human Genetics</i> , 2009, 125, 29-39.	3.8	139
3	The Developmental Pattern of Myotubes in Spinal Muscular Atrophy Indicates Prenatal Delay of Muscle Maturation. <i>Journal of Neuropathology and Experimental Neurology</i> , 2009, 68, 474-481.	1.7	80
4	Synaptic defects in type I spinal muscular atrophy in human development. <i>Journal of Pathology</i> , 2013, 229, 49-61.	4.5	77
5	The c.859G>C variant in the SMN2 gene is associated with types II and III SMA and originates from a common ancestor. <i>Journal of Medical Genetics</i> , 2010, 47, 640-642.	3.2	72
6	Plastin 3 expression in discordant spinal muscular atrophy (SMA) siblings. <i>Neuromuscular Disorders</i> , 2011, 21, 413-419.	0.6	52
7	Abnormalities in Early Markers of Muscle Involvement Support a Delay in Myogenesis in Spinal Muscular Atrophy. <i>Journal of Neuropathology and Experimental Neurology</i> , 2014, 73, 559-567.	1.7	36
8	Decay in survival motor neuron and plastin 3 levels during differentiation of iPSC-derived human motor neurons. <i>Scientific Reports</i> , 2015, 5, 11696.	3.3	32
9	Practical guidelines to manage discordant situations of <i>SMN2</i> copy number in patients with spinal muscular atrophy. <i>Neurology: Genetics</i> , 2020, 6, e530.	1.9	32
10	Accuracy of Marker Analysis, Quantitative Real-Time Polymerase Chain Reaction, and Multiple Ligation-Dependent Probe Amplification to Determine <i>SMN2</i> Copy Number in Patients with Spinal Muscular Atrophy. <i>Genetic Testing and Molecular Biomarkers</i> , 2011, 15, 587-594.	0.7	28
11	Utility of two SMN1 variants to improve spinal muscular atrophy carrier diagnosis and genetic counselling. <i>European Journal of Human Genetics</i> , 2018, 26, 1554-1557.	2.8	28
12	Treatment of spinal muscular atrophy cells with drugs that upregulate SMN expression reveals inter- and intra-patient variability. <i>European Journal of Human Genetics</i> , 2011, 19, 1059-1065.	2.8	25
13	Measurement of muscle strength with a handheld dynamometer in patients with chronic spinal muscular atrophy. <i>Journal of Rehabilitation Medicine</i> , 2010, 42, 228-231.	1.1	23
14	Beyond copy number: A new, rapid, and versatile method for sequencing the entire <i>SMN2</i> gene in SMA patients. <i>Human Mutation</i> , 2021, 42, 787-795.	2.5	23
15	A novel mutation in the caveolin-3 gene causing familial isolated hyperCKaemia. <i>Neuromuscular Disorders</i> , 2004, 14, 321-324.	0.6	16
16	Investigation of the role of SMN1 and SMN2 haploinsufficiency as a risk factor for Hirayama's disease: Clinical, neurophysiological and genetic characteristics in a Spanish series of 13 patients. <i>Clinical Neurology and Neurosurgery</i> , 2007, 109, 844-848.	1.4	15
17	Ultrasound evaluation of fetal movements in pregnancies at risk for severe spinal muscular atrophy. <i>Neuromuscular Disorders</i> , 2011, 21, 97-101.	0.6	15
18	Genotype-phenotype correlation of SMN locus genes in spinal muscular atrophy children from Argentina. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 910-917.	1.6	15

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19	High Mutational Heterogeneity, and New Mutations in the Human Coagulation Factor V Gene. Future Perspectives for Factor V Deficiency Using Recombinant and Advanced Therapies. International Journal of Molecular Sciences, 2021, 22, 9705.	4.1	9
20	Sarcomeric disorganization and nemaline bodies in muscle biopsies of patients with <i>EXOSC3</i> -related type 1 pontocerebellar hypoplasia. Muscle and Nerve, 2019, 59, 137-141.	2.2	8
21	Evidence of a segregation ratio distortion of SMN1 alleles in spinal muscular atrophy. European Journal of Human Genetics, 2007, 15, 1090-1093.	2.8	7
22	Evaluation of fetal nuchal translucency in 98 pregnancies at risk for severe spinal muscular atrophy: possible relevance of the SMN2 copy number. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 1246-1249.	1.5	7
23	Next-generation sequencing reveals a new mutation in the LTBP2 gene associated with microspherophakia in a Spanish family. BMC Medical Genetics, 2018, 19, 77.	2.1	5
24	Two independent mutations of the SMN1 gene in the same spinal muscular atrophy family branch: Lessons for carrier diagnosis. Genetics in Medicine, 2006, 8, 259-262.	2.4	2
25	Analysis of the <i>C9orf72</i> gene in spinal muscular atrophy patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 563-568.	1.7	2