Laura AlÃ-as

List of Publications by Year
in descending order

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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	Beyond copy number: A new, rapid, and versatile method for sequencing the entire SMN2 gene in SMA patients. Human Mutation, 2021, 42, 787-795.	2.5	23
2	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
3	Practical guidelines to manage discordant situations of SMN2 copy number in patients with spinal muscular atrophy. Neurology: Genetics, 2020, 6, e530.	1.9	32
4	Sarcomeric disorganization and nemaline bodies in muscle biopsies of patients with EXOSC3â€related type 1 pontocerebellar hypoplasia. Muscle and Nerve, 2019, 59, 137-141.	2.2	8
5	Correlation between SMA type and SMN2 copy number revisited: An analysis of 625 unrelated Spanish patients and a compilation of 2834 reported cases. Neuromuscular Disorders, 2018, 28, 208-215.	0.6	273
6	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
7	Utility of two SMN1 variants to improve spinal muscular atrophy carrier diagnosis and genetic counselling. European Journal of Human Genetics, 2018, 26, 1554-1557.	2.8	28
8	Genotypeâ€“phenotype correlation of SMN locus genes in spinal muscular atrophy children from Argentina. European Journal of Paediatric Neurology, 2016, 20, 910-917.	1.6	15
9	Decay in survival motor neuron and plastin 3 levels during differentiation of iPSC-derived human motor neurons. Scientific Reports, 2015, 5, 11696.	3.3	32
10	Abnormalities in Early Markers of Muscle Involvement Support a Delay in Myogenesis in Spinal Muscular Atrophy. Journal of Neuropathology and Experimental Neurology, 2014, 73, 559-567.	1.7	36
11	Analysis of the C9orf72 gene in spinal muscular atrophy patients. Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2014, 15, 563-568.	1.7	2
12	Synaptic defects in type I spinal muscular atrophy in human development. Journal of Pathology, 2013, 229, 49-61.	4.5	77
13	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
14	Ultrasound evaluation of fetal movements in pregnancies at risk for severe spinal muscular atrophy. Neuromuscular Disorders, 2011, 21, 97-101.	0.6	15
15	Plastin 3 expression in discordant spinal muscular atrophy (SMA) siblings. Neuromuscular Disorders, 2011, 21, 413-419.	0.6	52
16	Treatment of spinal muscular atrophy cells with drugs that upregulate SMN expression reveals inter- and intra-patient variability. European Journal of Human Genetics, 2011, 19, 1059-1065.	2.8	25
17	Accuracy of Marker Analysis, Quantitative Real-Time Polymerase Chain Reaction, and Multiple Ligation-Dependent Probe Amplification to Determine SMN2 Copy Number in Patients with Spinal Muscular Atrophy. Genetic Testing and Molecular Biomarkers, 2011, 15, 587-594.	0.7	28
18	Measurement of muscle strength with a handheld dynamometer in patients with chronic spinal muscular atrophy. Journal of Rehabilitation Medicine, 2010, 42, 228-231.	1.1	23

#	Article	IF	Citations
19	The c.859G>C variant in the SMN2 gene is associated with types II and III SMA and originates from a common ancestor. Journal of Medical Genetics, 2010, 47, 640-642.	3.2	72
20	Mutation update of spinal muscular atrophy in Spain: molecular characterization of 745 unrelated patients and identification of four novel mutations in the SMN1 gene. Human Genetics, 2009, 125, 29-39.	3.8	139
21	The Developmental Pattern of Myotubes in Spinal Muscular Atrophy Indicates Prenatal Delay of Muscle Maturation. Journal of Neuropathology and Experimental Neurology, 2009, 68, 474-481.	1.7	80
22	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. Clinical Neurology and Neurosurgery, 2007, 109, 844-848.	1.4	15
23	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
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	A novel mutation in the caveolin-3 gene causing familial isolated hyperCKaemia. Neuromuscular Disorders, 2004, 14, 321-324.	0.6	16