Sara Bernal

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/9301177/publications.pdf

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19	803	840776 11	839539 18
papers	citations	h-index	g-index

20	20	20	817
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Clinical and molecular response to dasatinib in an adult patient with Penttinen syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1233-1238.	1.2	1
2	Novel PLEKHG5 mutations in a patient with childhoodâ€onset lower motor neuron disease. Annals of Clinical and Translational Neurology, 2021, 8, 294-299.	3.7	2
3	Heterozygous APOE Christchurch in familial Alzheimerâ€™s disease without mutations in other Mendelian genes. Neuropathology and Applied Neurobiology, 2021, 47, 579-582.	3.2	10
4	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. Scientific Reports, 2021, 11, 1526.	3.3	71
5	Excellent response to secukinumab in an infant with severe generalized pustular psoriasis. Journal of Dermatology, 2021, 48, 907-910.	1.2	9
6	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
7	Association of the CFTR gene with asthma and airway mucus hypersecretion. PLoS ONE, 2021, 16, e0251881.	2.5	9
8	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
9	Frequency and clinical relevance of DPYD genetic variants in gastrointestinal cancer patients.. Farmacia Hospitalaria, 2021, 45, 5-10.	0.6	0
10	Practical guidelines to manage discordant situations of SMN2 copy number in patients with spinal muscular atrophy. Neurology: Genetics, 2020, 6, e530.	1.9	32
11	NHEJ-Mediated Repair of CRISPR-Cas9-Induced DNA Breaks Efficiently Corrects Mutations in HSPCs from Patients with Fanconi Anemia. Cell Stem Cell, 2019, 25, 607-621.e7.	11.1	64
12	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
13	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
14	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
15	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
16	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
17	Plastin 3 expression in discordant spinal muscular atrophy (SMA) siblings. Neuromuscular Disorders, 2011, 21, 413-419.	0.6	52
18	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

#	Article	IF	Citations
19	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