

# Sara Bernal

## List of Publications by Year in descending order

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

Version: 2024-02-01

19  
papers

803  
citations

840776

11  
h-index

839539

18  
g-index

20  
all docs

20  
docs citations

20  
times ranked

817  
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	Genetic landscape of 6089 inherited retinal dystrophies affected cases in Spain and their therapeutic and extended epidemiological implications. <i>Scientific Reports</i> , 2021, 11, 1526.	3.3	71
4	NHEJ-Mediated Repair of CRISPR-Cas9-Induced DNA Breaks Efficiently Corrects Mutations in HSPCs from Patients with Fanconi Anemia. <i>Cell Stem Cell</i> , 2019, 25, 607-621.e7.	11.1	64
5	Plastin 3 expression in discordant spinal muscular atrophy (SMA) siblings. <i>Neuromuscular Disorders</i> , 2011, 21, 413-419.	0.6	52
6	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
7	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
8	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
9	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
10	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
11	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
12	Heterozygous <i>APOE</i> Christchurch in familial Alzheimer's disease without mutations in other Mendelian genes. <i>Neuropathology and Applied Neurobiology</i> , 2021, 47, 579-582.	3.2	10
13	Excellent response to secukinumab in an infant with severe generalized pustular psoriasis. <i>Journal of Dermatology</i> , 2021, 48, 907-910.	1.2	9
14	Association of the CFTR gene with asthma and airway mucus hypersecretion. <i>PLoS ONE</i> , 2021, 16, e0251881.	2.5	9
15	High Mutational Heterogeneity, and New Mutations in the Human Coagulation Factor V Gene. Future Perspectives for Factor V Deficiency Using Recombinant and Advanced Therapies. <i>International Journal of Molecular Sciences</i> , 2021, 22, 9705.	4.1	9
16	Next-generation sequencing reveals a new mutation in the LTBP2 gene associated with microspherophakia in a Spanish family. <i>BMC Medical Genetics</i> , 2018, 19, 77.	2.1	5
17	Novel PLEKHG5 mutations in a patient with childhood-onset lower motor neuron disease. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 294-299.	3.7	2
18	Clinical and molecular response to dasatinib in an adult patient with Penttinen syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1233-1238.	1.2	1

#	ARTICLE	IF	CITATIONS
19	Frequency and clinical relevance of DPYD genetic variants in gastrointestinal cancer patients.. Farmacia Hospitalaria, 2021, 45, 5-10.	0.6	0