Jorge Alonso-PÃ©rez

List of Publications by Year
in descending order

26 papers	355 citations	1040056 9 h-index	888059 17 g-index
27 all docs	27 docs citations	27 times ranked	550 citing authors

#	Article	IF	Citations
1	Sequential targeted exome sequencing of 1001 patients affected by unexplained limb-girdle weakness. Genetics in Medicine, 2020, 22, 1478-1488.	2.4	62
2	New genotype-phenotype correlations in a large European cohort of patients with sarcoglycanopathy. Brain, 2020, 143, 2696-2708.	7.6	45
3	Accuracy of a machine learning muscle MRI-based tool for the diagnosis of muscular dystrophies. Neurology, 2020, 94, e1094-e1102.	1.1	45
4	Targeted Next-Generation Sequencing in a Large Cohort of Genetically Undiagnosed Patients with Neuromuscular Disorders in Spain. Genes, 2020, 11, 539.	2.4	25
5	Followâ€up of lateâ€onset Pompe disease patients with muscle magnetic resonance imaging reveals increase in fat replacement in skeletal muscles. Journal of Cachexia, Sarcopenia and Muscle, 2020, 11, 1032-1046.	7.3	25
6	STIG study: real-world data of long-term outcomes of adults with Pompe disease under enzyme replacement therapy with alglucosidase alfa. Journal of Neurology, 2021, 268, 2482-2492.	3.6	21
7	Growth Differentiation Factor 15 is a potential biomarker of therapeutic response for TK2 deficient myopathy. Scientific Reports, 2020, 10, 10111.	3.3	20
8	The increasing role of muscle MRI to monitor changes over time in untreated and treated muscle diseases. Current Opinion in Neurology, 2020, 33, 611-620.	3.6	18
9	Thrombospondin-1 mediates muscle damage in brachio-cervical inflammatory myopathy and systemic sclerosis. Neurology: Neuroimmunology and NeuroInflammation, 2020, 7, .	6.0	12
10	Clinical and genetic spectrum of a large cohort of patients with Î´-sarcoglycan muscular dystrophy. Brain, 2022, 145, 596-606.	7.6	11
11	Registro espaÃ±ol de la enfermedad de Pompe: anÃ¡lisis de los primeros 49 pacientes con enfermedad de Pompe del adulto. Medicina ClÃnica, 2020, 154, 80-85.	0.6	9
12	PDGF-BB serum levels are decreased in adult onset Pompe patients. Scientific Reports, 2019, 9, 2139.	3.3	8
13	Intramuscular fatty infiltration and physical function in controlled acromegaly. European Journal of Endocrinology, 2021, 185, 167-177.	3.7	7
14	Charcotâ€“Marieâ€“Tooth disease due to MORC2 mutations in Spain. European Journal of Neurology, 2021, 28, 3001-3011.	3.3	6
15	Late onset Sandhoff disease presenting with lower motor neuron disease and stuttering. Neuromuscular Disorders, 2021, 31, 769-772.	0.6	6
16	Study of the effect of anti-rhGAA antibodies at low and intermediate titers in late onset Pompe patients treated with ERT. Molecular Genetics and Metabolism, 2019, 128, 129-136.	1.1	5
17	Correlation Between Respiratory Accessory Muscles and Diaphragm Pillars MRI and Pulmonary Function Test in Late-Onset Pompe Disease Patients. Frontiers in Neurology, 2021, 12, 621257.	2.4	5
18	Magnetization Transfer Ratio in Lower Limbs of Late Onset Pompe Patients Correlates With Intramuscular Fat Fraction and Muscle Function Tests. Frontiers in Neurology, 2021, 12, 634766.	2.4	4

#	Article	IF	Citations
19	Clinical and genetic features of a large homogeneous cohort of oculopharyngeal muscular dystrophy patients from the Canary Islands. European Journal of Neurology, 2022, 29, 1488-1495.	3.3	4
20	Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis. Journal of Neurology, 2022, 269, 3550-3562.	3.6	4
21	Spanish Pompe registry: Baseline characteristics of first 49 patients with adult onset of Pompe disease. Medicina ClÃnica (English Edition), 2020, 154, 80-85.	0.2	3
22	Platelet Derived Growth Factor-AA Correlates With Muscle Function Tests and Quantitative Muscle Magnetic Resonance in Dystrophinopathies. Frontiers in Neurology, 2021, 12, 659922.	2.4	3
23	Different Approaches to Analyze Muscle Fat Replacement With Dixon MRI in Pompe Disease. Frontiers in Neurology, 2021, 12, 675781.	2.4	3
24	Isolation of human fibroadipogenic progenitors and satellite cells from frozen muscle biopsies. FASEB Journal, 2021, 35, e21819.	0.5	3
25	An Integrative Analysis of DNA Methylation Pattern in Myotonic Dystrophy Type 1 Samples Reveals a Distinct DNA Methylation Profile between Tissues and a Novel Muscle-Associated Epigenetic Dysregulation. Biomedicines, 2022, 10, 1372.	3.2	1
26	High prevalence of paraspinal muscle involvement in adults with <scp>McArdle</scp> disease. Muscle and Nerve, 2022, , .	2.2	0