Teresinha Evangelista

List of Publications by Year
in descending order

69	2,150	186265	243625
papers	citations	28 h-index	44 g-index

74	74	74	3291
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Development of a standard of care for patients with valosin-containing protein associated multisystem proteinopathy. Orphanet Journal of Rare Diseases, 2022, 17, 23.	2.7	19
2	Convergence of patient- and physician-reported outcomes in the French National Registry of Facioscapulohumeral Dystrophy. Orphanet Journal of Rare Diseases, 2022, 17, 96.	2.7	2
3	Heterozygous frameshift variants in HNRNPA2B1 cause early-onset oculopharyngeal muscular dystrophy. Nature Communications, 2022, 13, 2306.	12.8	20
4	Survey on patientsâ€™ organisationsâ€™ knowledge and position paper on screening for inherited neuromuscular diseases in Europe. Orphanet Journal of Rare Diseases, 2021, 16, 75.	2.7	3
5	Genetic testing offer for inherited neuromuscular diseases within the EURO-NMD reference network: A European survey study. PLoS ONE, 2020, 15, e0239329.	2.5	6
6	Facioscapulohumeral muscular dystrophy 1 patients participating in the UK FSHD registry can be subdivided into 4 patterns of self-reported symptoms. Neuromuscular Disorders, 2020, 30, 315-328.	0.6	15
7	Title is missing!. , 2020, 15, e0239329.		0
8	Title is missing!. , 2020, 15, e0239329.		0
9	Title is missing!. , 2020, 15, e0239329.		0
10	Title is missing!. , 2020, 15, e0239329.		0
11	SMCHD1 mutation spectrum for facioscapulohumeral muscular dystrophy type 2 (FSHD2) and Bosma arhinia microphthalmia syndrome (BAMS) reveals disease-specific localisation of variants in the ATPase domain. Journal of Medical Genetics, 2019, 56, 693-700.	3.2	27
12	HNRNPDL-related muscular dystrophy: expanding the clinical, morphological and MRI phenotypes. Journal of Neurology, 2019, 266, 2524-2534.	3.6	18
13	Correction of pseudoexon splicing caused by a novel intronic dysferlin mutation. Annals of Clinical and Translational Neurology, 2019, 6, 642-654.	3.7	20
14	Phenotype may predict the clinical course of facioscapolohumeral muscular dystrophy. Muscle and Nerve, 2019, 59, 711-713.	2.2	12
15	237th ENMC International Workshop: GNE myopathy â€“ current and future research Hoofddorp, The Netherlands, 14â€“16 September 2018. Neuromuscular Disorders, 2019, 29, 401-410.	0.6	5
16	Recessive variants of MuSK are associated with late onset CMS and predominant limb girdle weakness. American Journal of Medical Genetics, Part A, 2018, 176, 1594-1601.	1.2	25
17	Whole-exome sequencing identifies mutations in MYMK in a mild form of Carey-Fineman-Ziter syndrome. Neurology: Genetics, 2018, 4, e226.	1.9	6
18	Chronic pain has a strong impact on quality of life in facioscapulohumeral muscular dystrophy. Muscle and Nerve, 2018, 57, 380-387.	2.2	33

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19	Congenital myasthenic syndrome with episodic apnoea: clinical, neurophysiological and genetic features in the long-term follow-up of 19 patients. Journal of Neurology, 2018, 265, 194-203.	3.6	36
20	FSHD type 2 and Bosma arhinia microphthalmia syndrome. Neurology, 2018, 91, e562-e570.	1.1	24
21	Enzyme replacement therapy with alglucosidase alfa in a late-onset Pompe disease patient during pregnancy. Neuromuscular Disorders, 2018, 28, 965-968.	0.6	4
22	Reduced serum myostatin concentrations associated with genetic muscle disease progression. Journal of Neurology, 2017, 264, 541-553.	3.6	51
23	How the EUCERD Joint Action supported initiatives on Rare Diseases. European Journal of Medical Genetics, 2017, 60, 185-189.	1.3	7
24	219th ENMC International Workshop Titinopathies International database of titin mutations and phenotypes, Heemskerk, The Netherlands, 29 Aprilâ€“1 May 2016. Neuromuscular Disorders, 2017, 27, 396-407.	0.6	29
25	Genetic heterogeneity of motor neuropathies. Neurology, 2017, 88, 1226-1234.	1.1	81
26	225th ENMC international workshop:. Neuromuscular Disorders, 2017, 27, 782-790.	0.6	20
27	Screening for Pompe disease in a Portuguese high risk population. Neuromuscular Disorders, 2017, 27, 777-781.	0.6	12
28	Intragenic DOK7 deletion detected by whole-genome sequencing in congenital myasthenic syndromes. Neurology: Genetics, 2017, 3, e152.	1.9	4
29	Complex phenotypes associated with STIM1 mutations in both coiled coil and EF-hand domains. Neuromuscular Disorders, 2017, 27, 861-872.	0.6	39
30	The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. Neuropediatrics, 2017, 48, 294-308.	0.6	43
31	Respiratory involvement in ambulant and non-ambulant patients with facioscapulohumeral muscular dystrophy. Journal of Neurology, 2017, 264, 1271-1280.	3.6	30
32	Molecular characterization of congenital myasthenic syndromes in Spain. Neuromuscular Disorders, 2017, 27, 1087-1098.	0.6	51
33	Co-presentation of adult-onset systemic lupus erythematosus and nemaline myopathy. Rheumatology, 2017, 56, 2034-2035.	1.9	5
34	Cytokine Profiling of Serum Allows Monitoring of Disease Progression in Inclusion Body Myositis. Journal of Neuromuscular Diseases, 2017, 4, 327-335.	2.6	8
35	Exome sequences versus sequential gene testing in the UK highly specialised Service for Limb Girdle Muscular Dystrophy. Orphanet Journal of Rare Diseases, 2017, 12, 151.	2.7	44
36	Prevalence of Pompe disease in 3,076 patients with hyperCKemia and limb-girdle muscular weakness. Neurology, 2016, 87, 295-298.	1.1	60

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37	Design, set-up and utility of the UK facioscapulohumeral muscular dystrophy patient registry. Journal of Neurology, 2016, 263, 1401-1408.	3.6	25
38	215th ENMC International Workshop VCP-related multi-system proteinopathy (IBMPFD) 13â€“15 November 2015, Heemskerk, The Netherlands. Neuromuscular Disorders, 2016, 26, 535-547.	0.6	38
39	The context for the thematic grouping of rare diseases to facilitate the establishment of European Reference Networks. Orphanet Journal of Rare Diseases, 2016, 11, 17.	2.7	31
40	Identification of mutations in theMYO9Agene in patients with congenital myasthenic syndrome. Brain, 2016, 139, 2143-2153.	7.6	45
41	Long-term follow-up in patients with congenital myasthenic syndrome due to RAPSN mutations. Neuromuscular Disorders, 2016, 26, 153-159.	0.6	31
42	Congenital Myasthenic Syndromes with Predominant Limb Girdle Weakness. Journal of Neuromuscular Diseases, 2015, 2, S21-S29.	2.6	28
43	Phenotypic variability of TRPV4 related neuropathies. Neuromuscular Disorders, 2015, 25, 516-521.	0.6	27
44	Two recurrent mutations are associated with GNE myopathy in the North of Britain. Journal of Neurology, Neurosurgery and Psychiatry, 2014, 85, 1359-1365.	1.9	30
45	Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. Brain, 2014, 137, 2429-2443.	7.6	86
46	Atypical phenotype in two patients with LAMA2 mutations. Neuromuscular Disorders, 2014, 24, 419-424.	0.6	30
47	New variants, challenges and pitfalls in DMD genotyping: implications in diagnosis, prognosis and therapy. Journal of Human Genetics, 2014, 59, 454-464.	2.3	35
48	Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Journal of Neuromuscular Diseases, 2014, 1, 75-90.	2.6	69
49	Centres of Expertise and European Reference Networks: key issues in the field of rare diseases. The EUCERD Recommendations. Blood Transfusion, 2014, 12 Suppl 3, s621-5.	0.4	14
50	â€œDouble-troubleâ€•or digenic disorder in complex I deficiency. Mitochondrion, 2012, 12, 585.	3.4	0
51	Liver and Muscle in Morbid Obesity: The Interplay of Fatty Liver and Insulin Resistance. PLoS ONE, 2012, 7, e31738.	2.5	38
52	Relative frequency of known causes of multiple mtDNA deletions: Two novel POLG mutations. Neuromuscular Disorders, 2011, 21, 483-488.	0.6	16
53	Dominant and recessive RYR1 mutations in adults with core lesions and mild muscle symptoms. Muscle and Nerve, 2011, 44, 102-108.	2.2	39
54	Acquired amyloid neuropathy in a Portuguese patient after domino liver transplantation. Muscle and Nerve, 2010, 42, 836-838.	2.2	25

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55	Private dysferlin exon skipping mutation (c.5492G>A) with a founder effect reveals further alternative splicing involving exons 49â€“51. Journal of Human Genetics, 2010, 55, 546-549.	2.3	12
56	A randomized, placebo-controlled trial of memantine for functional disability in amyotrophic lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2010, 11, 456-460.	2.1	101
57	A case of asymptomatic cytoplasmic body myopathy revealed by sinvastatin. Neuromuscular Disorders, 2009, 19, 66-68.	0.6	5
58	Botulinum toxin type-B improves sialorrhea and quality of life in bulbaronset amyotrophic lateral sclerosis. Journal of Neurology, 2008, 255, 545-550.	3.6	51
59	Sleep characteristics of amyotrophic lateral sclerosis in patients with preserved diaphragmatic function. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2007, 8, 101-105.	2.1	75
60	A new mtDNAâ€tRNAGlu mutation (14728T>C) presenting a late-onset mitochondrial encephalomyopathy. Mitochondrion, 2007, 7, 396-398.	3.4	8
61	Evidence for central abnormality in respiratory control in primary lateral sclerosis. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders, 2006, 7, 57-60.	2.1	7
62	Abrupt onset of progressive muscular atrophy. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2004, 5, 61-62.	1.2	0
63	Nocturnal pulse oximetry: a new approach to establish the appropriate time for nonâ€invasive ventilation in ALS patients. Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases, 2003, 4, 31-35.	1.2	95
64	New transthyretin mutation V28M in a Portuguese kindred with amyloid polyneuropathy. Muscle and Nerve, 2000, 23, 1016-1021.	2.2	18
65	Respiratory disorders in ALS: sleep and exercise studies. Journal of the Neurological Sciences, 1999, 169, 61-68.	0.6	29
66	Can amyotrophic lateral sclerosis patients with respiratory insufficiency exercise?. Journal of the Neurological Sciences, 1999, 169, 69-75.	0.6	88
67	Motor neuropathies mimicking amyotrophic lateral sclerosis/motor neuron disease. Journal of the Neurological Sciences, 1996, 139, 95-98.	0.6	32
68	Phrenic nerve conduction in amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 1995, 129, 35-37.	0.6	45
69	Localization of merosin-negative congenital muscular dystrophy to chromosome 6q2 by homozygosity mapping. Human Molecular Genetics, 1994, 3, 1657-1661.	2.9	214