Montse Olive

List of Publications by Year
in descending order

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123	4,074	94381	143943
papers	citations	37 h-index	57 g-index

132	132	132	4801
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	High prevalence of paraspinal muscle involvement in adults with <scp>McArdle</scp> disease. Muscle and Nerve, 2022, , .	1.0	0
2	Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis. Journal of Neurology, 2022, 269, 3550-3562.	1.8	4
3	BNIP3 Is Involved in Muscle Fiber Atrophy in Late-Onset Pompe Disease Patients. American Journal of Pathology, 2022, , .	1.9	3
4	Immune Response and Safety of SARS-CoV-2 mRNA-1273 Vaccine in Patients With Myasthenia Gravis. Neurology: Neuroimmunology and NeuroInflammation, 2022, 9, .	3.1	15
5	246th ENMC International Workshop: Protein aggregate myopathies 24â€“26 May 2019, Hoofddorp, The Netherlands. Neuromuscular Disorders, 2021, 31, 158-166.	0.3	5
6	Heterozygous CAPN3 missense variants causing autosomalâ€dominant calpainopathy in seven unrelated families. Neuropathology and Applied Neurobiology, 2021, 47, 283-296.	1.8	23
7	Association of Initial Maximal Motor Ability With Long-term Functional Outcome in Patients With COL6-Related Dystrophies. Neurology, 2021, 96, e1413-e1424.	1.5	10
8	Collaborative model for diagnosis and treatment of very rare diseases: experience in Spain with thymidine kinase 2 deficiency. Orphanet Journal of Rare Diseases, 2021, 16, 407.	1.2	3
9	Mutations in the J domain of DNAJB6 cause dominant distal myopathy. Neuromuscular Disorders, 2020, 30, 38-46.	0.3	20
10	Cylindrical spirals in two families: Clinical and genetic investigations. Neuromuscular Disorders, 2020, 30, 151-158.	0.3	7
11	ASCâ€1 Is a Cell Cycle Regulator Associated with Severe and Mild Forms of Myopathy. Annals of Neurology, 2020, 87, 217-232.	2.8	12
12	Impaired muscle morphology in aDrosophilamodel of myosin storage myopathy was supressed by overexpression of an E3 ubiquitin ligase. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	2
13	Targeted Next-Generation Sequencing in a Large Cohort of Genetically Undiagnosed Patients with Neuromuscular Disorders in Spain. Genes, 2020, 11, 539.	1.0	25
14	HNRNPDL-related limb girdle muscular dystrophy in a Spanish family with scapulo-peroneal phenotype, the first family in Europe. Journal of the Neurological Sciences, 2020, 414, 116875.	0.3	2
15	Mutations in TRIM63 cause an autosomal-recessive form of hypertrophic cardiomyopathy. Heart, 2020, 106, 1342-1348.	1.2	27
16	SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e75-e76.	1.7	1
17	Spanish Pompe registry: Baseline characteristics of first 49 patients with adult onset of Pompe disease. Medicina ClÃnica (English Edition), 2020, 154, 80-85.	0.1	3
18	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.	0.3	9

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19	Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. Brain, 2019, 142, e66-e66.	3.7	8
20	Motor neuron diseases caused by a novel VRK1 variant â€“ A genotype/phenotype study. Annals of Clinical and Translational Neurology, 2019, 6, 2197-2204.	1.7	12
21	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	1.2	29
22	Actininopathy: A new muscular dystrophy caused by ACTN2 dominant mutations. Annals of Neurology, 2019, 85, 899-906.	2.8	22
23	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	5.8	11
24	1st ENMC European meeting: The EURO-NMD pathology working group Recommended Standards for Muscle Pathology Amsterdam, The Netherlands, 7 December 2018. Neuromuscular Disorders, 2019, 29, 483-485.	0.3	27
25	Late onset distal myopathy: A new telethoninopathy. Neuromuscular Disorders, 2019, 29, 80-83.	0.3	0
26	Muscle MRI in a large cohort of patients with oculopharyngeal muscular dystrophy. Journal of Neurology, Neurosurgery and Psychiatry, 2019, 90, 576-585.	0.9	38
27	Ryanodine receptor type 3 (<scp>RYR</scp>3) as a novel gene associated with a myopathy with nemaline bodies. European Journal of Neurology, 2018, 25, 841-847.	1.7	31
28	Dilated Cardiomyopathy and Hydroxychloroquine-induced Phospholipidosis: From Curvilinear Bodies to Clinical Suspicion. Revista Espanola De Cardiologia (English Ed), 2018, 71, 491-493.	0.4	4
29	MiocardiopatÃa dilatada y fosfolipidosis inducida por hidroxicloroquina: de los cuerpos curvilÃneos a la sospecha clÃnica. Revista Espanola De Cardiologia, 2018, 71, 491-493.	0.6	5
30	CONGENITAL MUSCULAR DYSTROPHIES. Neuromuscular Disorders, 2018, 28, S130.	0.3	0
31	Drosophila model of myosin myopathy rescued by overexpression of a TRIM-protein family member. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E6566-E6575.	3.3	10
32	A Roma founder BIN1 mutation causes a novel phenotype of centronuclear myopathy with rigid spine. Neurology, 2018, 91, e339-e348.	1.5	18
33	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for followâ€up. Muscle and Nerve, 2018, 58, 812-817.	1.0	8
34	Non-compaction cardiomyopathy and early respiratory failure in an adult symptomatic female carrier of centronuclear myopathy caused by a MTM1 mutation. Neuromuscular Disorders, 2018, 28, 952-955.	0.3	7
35	Different mitochondrial genetic defects exhibit the same protein signature of metabolism in skeletal muscle of PEO and MELAS patients: A role for oxidative stress. Free Radical Biology and Medicine, 2018, 126, 235-248.	1.3	10
36	Molecular characterization of congenital myasthenic syndromes in Spain. Neuromuscular Disorders, 2017, 27, 1087-1098.	0.3	51

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37	Hypoxia triggers IFN-I production in muscle: Implications in dermatomyositis. Scientific Reports, 2017, 7, 8595.	1.6	30
38	BIN1 founder mutation in the Spanish gypsy population is the most frequent cause of adult onset centronuclear myopathies in the south of Spain. Neuromuscular Disorders, 2017, 27, S172-S173.	0.3	0
39	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	1.9	38
40	Differences in Adipose Tissue and Lean Mass Distribution in Patients with Collagen VI Related Myopathies Are Associated with Disease Severity and Physical Ability. Frontiers in Aging Neuroscience, 2017, 9, 268.	1.7	7
41	Cutaneous Neoplasms in Myotonic Dystrophy Type 1. Dermatology, 2016, 232, 700-703.	0.9	10
42	New aspects of myofibrillar myopathies. Current Opinion in Neurology, 2016, 29, 628-634.	1.8	57
43	Ryanodine receptor type 3 (RYR3) as a novel gene associated with nemaline myopathy and fibre type disproportion. Neuromuscular Disorders, 2016, 26, S137.	0.3	3
44	Pediatric onset of mitochondrial myopathy due to ANT1 mutation. Neuromuscular Disorders, 2016, 26, S176.	0.3	0
45	Novel mutation in TCAP manifesting with asymmetric calves and early-onset joint retractions. Neuromuscular Disorders, 2016, 26, 749-753.	0.3	16
46	New insights into the protein aggregation pathology in myotilinopathy by combined proteomic and immunolocalization analyses. Acta Neuropathologica Communications, 2016, 4, 8.	2.4	50
47	Muscle imaging in muscle dystrophies produced by mutations in the EMD and LMNA genes. Neuromuscular Disorders, 2016, 26, 33-40.	0.3	40
48	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. PLoS ONE, 2016, 11, e0156359.	1.1	48
49	Increased amyloid Î²-peptide uptake in skeletal muscle is induced by hyposialylation and may account for apoptosis in GNE myopathy. Oncotarget, 2016, 7, 13354-13371.	0.8	16
50	Dried Blood Spot for Screening for Late-Onset Pompe Disease: A Spanish Cohort. Journal of Neuromuscular Diseases, 2015, 2, S42-S42.	1.1	3
51	Targeted screening for the detection of Pompe disease in patients with unclassified limb-girdle muscular dystrophy or asymptomatic hyperCKemia using dried blood: A Spanish cohort. Neuromuscular Disorders, 2015, 25, 548-553.	0.3	47
52	New cardiac and skeletal protein aggregate myopathy associated with combined MuRF1 and MuRF3 mutations. Human Molecular Genetics, 2015, 24, 3638-3650.	1.4	51
53	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. Brain, 2014, 137, 3160-3170.	3.7	76
54	G.P.38. Neuromuscular Disorders, 2014, 24, 806.	0.3	0

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55	A new disease allele for the p.C30071R mutation in titin causing hereditary myopathy with early respiratory failure. Neuromuscular Disorders, 2014, 24, 241-244.	0.3	10
56	Exome sequencing identifies titin mutations causing hereditary myopathy with early respiratory failure (HMERF) in families of diverse ethnic origins. BMC Neurology, 2013, 13, 29.	0.8	37
57	Filamin C-related myopathies: pathology and mechanisms. Acta Neuropathologica, 2013, 125, 33-46.	3.9	106
58	Differential proteomic analysis of abnormal intramyoplasmic aggregates in desminopathy. Journal of Proteomics, 2013, 90, 14-27.	1.2	63
59	Skeletal Muscle Biopsy Analysis in Reducing Body Myopathy and Other FHL1-Related Disorders. Journal of Neuropathology and Experimental Neurology, 2013, 72, 833-845.	0.9	36
60	Myofibrillar myopathies. Current Opinion in Neurology, 2013, 26, 527-535.	1.8	48
61	Impairment of protein degradation in myofibrillar myopathy caused by FLNC/filamin C mutations. Autophagy, 2013, 9, 422-423.	4.3	30
62	Myofibrillar Myopathies. , 2013, , 247-265.		1
63	Pathophysiology of protein aggregation and extended phenotyping in filaminopathy. Brain, 2012, 135, 2642-2660.	3.7	70
64	Prognostic value of X-chromosome inactivation in symptomatic female carriers of dystrophinopathy. Orphanet Journal of Rare Diseases, 2012, 7, 82.	1.2	40
65	Isolated cardiomyopathy caused by a DMD nonsense mutation in somatic mosaicism: genetic normalization in skeletal muscle. Clinical Genetics, 2012, 82, 574-578.	1.0	10
66	Telethonin-deficiency initially presenting as a congenital muscular dystrophy. Neuromuscular Disorders, 2011, 21, 433-438.	0.3	37
67	Clinical and myopathological evaluation of early- and late-onset subtypes of myofibrillar myopathy. Neuromuscular Disorders, 2011, 21, 533-542.	0.3	135
68	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2011, 88, 122.	2.6	0
69	Dominant Mutations in KBTBD13, a Member of the BTB/Kelch Family, Cause Nemaline Myopathy with Cores. American Journal of Human Genetics, 2010, 87, 842-847.	2.6	143
70	Nemaline myopathy type 6: Clinical and myopathological features. Muscle and Nerve, 2010, 42, 901-907.	1.0	19
71	P1.48 Unusual morphological changes due to different mutations in lamin A/C gene: observations in four patients. Neuromuscular Disorders, 2010, 20, 615.	0.3	0
72	Glycogen branching enzyme deficiency in an infant with severe congenital hypotonia: an emerging diagnosis of muscle weakness in the perinatal period. Histopathology, 2009, 54, 765-768.	1.6	7

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73	In-frame deletion in the seventh immunoglobulin-like repeat of filamin C in a family with myofibrillar myopathy. European Journal of Human Genetics, 2009, 17, 656-663.	1.4	71
74	Extralysosomal Protein Degradation in Myofibrillar Myopathies. Brain Pathology, 2009, 19, 507-515.	2.1	16
75	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. Neuromuscular Disorders, 2009, 19, 418-422.	0.3	58
76	Generalized muscle pseudo-hypertrophy and stiffness associated with the myotilin Ser55Phe mutation: A novel myotilinopathy phenotype?. Journal of the Neurological Sciences, 2009, 277, 167-171.	0.3	14
77	TAR DNA-Binding Protein 43 Accumulation in Protein Aggregate Myopathies. Journal of Neuropathology and Experimental Neurology, 2009, 68, 262-273.	0.9	83
78	Expression of mutant ubiquitin (UBB⁺¹) and p62 in myotilinopathies and desminopathies. Neuropathology and Applied Neurobiology, 2008, 34, 76-87.	1.8	48
79	156th ENMC International Workshop: Desmin and protein aggregate myopathies, 9â€“11 November 2007, Naarden, The Netherlands. Neuromuscular Disorders, 2008, 18, 583-592.	0.3	15
80	Transcription-terminating mutation in telethonin causing autosomal recessive muscular dystrophy type 2G in a European patient. Neuromuscular Disorders, 2008, 18, 929-933.	0.3	48
81	Molecular pathology of myofibrillar myopathies. Expert Reviews in Molecular Medicine, 2008, 10, e25.	1.6	67
82	Distinct muscle imaging patterns in myofibrillar myopathies. Neurology, 2008, 71, 758-765.	1.5	204
83	Intermediate Filament Diseases: Desminopathy. Advances in Experimental Medicine and Biology, 2008, 642, 131-164.	0.8	97
84	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.5	120
85	Desmin Is Oxidized and Nitrated in Affected Muscles in Myotilinopathies and Desminopathies. Journal of Neuropathology and Experimental Neurology, 2007, 66, 711-723.	0.9	33
86	Phenotypic patterns of desminopathy associated with three novel mutations in the desmin gene. Neuromuscular Disorders, 2007, 17, 443-450.	0.3	50
87	Target Genes of Neuron-Restrictive Silencer Factor Are Abnormally Up-Regulated in Human Myotilinopathy. American Journal of Pathology, 2007, 171, 1312-1323.	1.9	30
88	Oxidative Stress in Desminopathies and Myotilinopathies: A Link between Oxidative Damage and Abnormal Protein Aggregation. Brain Pathology, 2007, 17, 377-388.	2.1	43
89	Different early pathogenesis in myotilinopathy compared to primary desminopathy. Neuromuscular Disorders, 2006, 16, 361-367.	0.3	29
90	Involvement of Clusterin and the Aggresome in Abnormal Protein Deposits in Myofibrillar Myopathies and Inclusion Body Myositis. Brain Pathology, 2006, 15, 101-108.	2.1	36

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91	NARP-MILS syndrome caused by 8993 TÂ>ÂG mitochondrial DNA mutation: a clinical, genetic and neuropathological study. Acta Neuropathologica, 2006, 111, 610-616.	3.9	47
92	Myotilinopathy: refining the clinical and myopathological phenotype. Brain, 2005, 128, 2315-2326.	3.7	146
93	A novel thymidine phosphorylase mutation in a Spanish MNGIE patient. Journal of the Neurological Sciences, 2005, 228, 35-39.	0.3	20
94	Small deletions disturb desmin architecture leading to breakdown of muscle cells and development of skeletal or cardioskeletal myopathy. Human Genetics, 2004, 114, 306-313.	1.8	59
95	A series of West European patients with severe cardiac and skeletal myopathy associated with a de novo R406W mutation in desmin. Journal of Neurology, 2004, 251, 143-149.	1.8	53
96	Overexpression of semicarbazide-sensitive amine oxidase in human myopathies. Muscle and Nerve, 2004, 29, 261-266.	1.0	13
97	Desmin-related myopathy: clinical, electrophysiological, radiological, neuropathological and genetic studies. Journal of the Neurological Sciences, 2004, 219, 125-137.	0.3	72
98	A novel mutation in the caveolin-3 gene causing familial isolated hyperCKaemia. Neuromuscular Disorders, 2004, 14, 321-324.	0.3	16
99	Proteasomal Expression, Induction of Immunoproteasome Subunits, and Local MHC Class I Presentation in Myofibrillar Myopathy and Inclusion Body Myositis. Journal of Neuropathology and Experimental Neurology, 2004, 63, 484-498.	0.9	84
100	Expression of the intermediate filament protein synemin in myofibrillar myopathies and other muscle diseases. Acta Neuropathologica, 2003, 106, 1-7.	3.9	29
101	Connexin 31 (GJB3) is expressed in the peripheral and auditory nerves and causes neuropathy and hearing impairment. Human Molecular Genetics, 2001, 10, 947-952.	1.4	109
102	Diabetic muscular infarction. , 2000, 23, 825-826.		10
103	Bcl-2 and Bax immunohistochemistry in denervation-reinnervation and necrosis-regeneration of rat skeletal muscles. Muscle and Nerve, 2000, 23, 1862-1867.	1.0	22
104	Bcl-2 and Bax protein expression in human myopathies. Journal of the Neurological Sciences, 1999, 164, 76-81.	0.3	32
105	Expression of myogenic regulatory factors (MRFs) in human neuromuscular disorders. Neuropathology and Applied Neurobiology, 1997, 23, 475-482.	1.8	22
106	Apoptosis is not the mechanism of cell death of muscle fibers in human muscular dystrophies and inflammatory myopathies. , 1997, 20, 1328-1330.		33
107	Expression of myogenic regulatory factors (MRFs) in human neuromuscular disorders. Neuropathology and Applied Neurobiology, 1997, 23, 475-482.	1.8	3
108	Strong c-Jun immunoreactivity is associated with apoptotic cell death in human tumors of the central nervous system. Neuroscience Letters, 1996, 214, 49-52.	1.0	7

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109	Selective c-Jun overexpression is associated with ionizing radiation-induced apoptosis in the developing cerebellum of the rat. Molecular Brain Research, 1996, 38, 91-100.	2.5	50
110	Transforming growth factor-Î± (TGF-Î±) and epidermal growth factor-receptor (EGF-R) immunoreactivity in normal and pathologic brain. Progress in Neurobiology, 1996, 49, 99-119.	2.8	85
111	CREB-1 and CREB-2 immunoreactivity in the rat brain. Brain Research, 1996, 712, 159-164.	1.1	16
112	Naturally Occurring (Programmed) and Radiation-induced Apoptosis are Associated with Selective c-Jun Expression in the Developing Rat Brain. European Journal of Neuroscience, 1996, 8, 1286-1298.	1.2	77
113	Strong c-Jun immunoreactivity is associated with apoptotic cell death in human tumors of the central nervous system. Neuroscience Letters, 1996, 214, 49-52.	1.0	30
114	Hereditary spastic paraparesis with dementia, amyotrophy and peripheral neuropathy. A neuropathological study. Neuropathology and Applied Neurobiology, 1995, 21, 255-261.	1.8	30
115	Amoeboid microglial response following X-ray-induced apoptosis in the neonatal rat brain. Neuroscience Letters, 1995, 193, 109-112.	1.0	13
116	Evidence of internucleosomal DNA fragmentation and identification of dying cells in X-ray-induced cell death in the developing brain. International Journal of Developmental Neuroscience, 1995, 13, 21-28.	0.7	32
117	Transforming growth factor-Î± immunoreactivity in the developing and adult brain. Neuroscience, 1995, 66, 189-199.	1.1	73
118	Cell death induced by gamma irradiation of developing skeletal muscle. Journal of Anatomy, 1995, 187 () Tj ETQq0 0 0 rgBT /Overlock 10	0.9	7
119	Congenital Muscular Dystrophy with Distinct CNS Involvement. Neuropediatrics, 1994, 25, 48-50.	0.3	2
120	Postnatal development of parvalbumin immunoreactivity in striated muscles of the rat. Anatomy and Embryology, 1994, 190, 301-5.	1.5	4
121	Parvalbumin immunocytochemistry and calcium deposition in muscle fiber necrosis and subsequent regeneration following intramuscular injection of metoclopramide. Muscle and Nerve, 1994, 17, 494-499.	1.0	9
122	Parvalbumin immunohistochemistry in denervated skeletal muscle. Neuropathology and Applied Neurobiology, 1994, 20, 495-500.	1.8	12
123	Radiosensitive populations and recovery in X-ray-induced apoptosis in the developing cerebellum. Acta Neuropathologica, 1993, 86, 491-500.	3.9	35