Montse Olive

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/347924/publications.pdf

Version: 2024-02-01

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	Distinct muscle imaging patterns in myofibrillar myopathies. Neurology, 2008, 71, 758-765.	1.5	204
2	Myotilinopathy: refining the clinical and myopathological phenotype. Brain, 2005, 128, 2315-2326.	3.7	146
3	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
4	Clinical and myopathological evaluation of early- and late-onset subtypes of myofibrillar myopathy. Neuromuscular Disorders, 2011, 21, 533-542.	0.3	135
5	Phenotypic clustering of lamin A/C mutations in neuromuscular patients. Neurology, 2007, 69, 1285-1292.	1.5	120
6	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
7	Filamin C-related myopathies: pathology and mechanisms. Acta Neuropathologica, 2013, 125, 33-46.	3.9	106
8	Intermediate Filament Diseases: Desminopathy. Advances in Experimental Medicine and Biology, 2008, 642, 131-164.	0.8	97
9	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
10	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
11	TAR DNA-Binding Protein 43 Accumulation in Protein Aggregate Myopathies. Journal of Neuropathology and Experimental Neurology, 2009, 68, 262-273.	0.9	83
12	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
13	Adult-onset autosomal dominant centronuclear myopathy due to BIN1 mutations. Brain, 2014, 137, 3160-3170.	3.7	76
14	Transforming growth factor-Î± immunoreactivity in the developing and adult brain. Neuroscience, 1995, 66, 189-199.	1.1	73
15	Desmin-related myopathy: clinical, electrophysiological, radiological, neuropathological and genetic studies. Journal of the Neurological Sciences, 2004, 219, 125-137.	0.3	72
16	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
17	Pathophysiology of protein aggregation and extended phenotyping in filaminopathy. Brain, 2012, 135, 2642-2660.	3.7	70
18	Molecular pathology of myofibrillar myopathies. Expert Reviews in Molecular Medicine, 2008, 10, e25.	1.6	67

#	Article	IF	Citations
19	Differential proteomic analysis of abnormal intramyoplasmic aggregates in desminopathy. Journal of Proteomics, 2013, 90, 14-27.	1.2	63
20	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
21	Severe infantile-onset cardiomyopathy associated with a homozygous deletion in desmin. Neuromuscular Disorders, 2009, 19, 418-422.	0.3	58
22	New aspects of myofibrillar myopathies. Current Opinion in Neurology, 2016, 29, 628-634.	1.8	57
23	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
24	New cardiac and skeletal protein aggregate myopathy associated with combined MuRF1 and MuRF3 mutations. Human Molecular Genetics, 2015, 24, 3638-3650.	1.4	51
25	Molecular characterization of congenital myasthenic syndromes in Spain. Neuromuscular Disorders, 2017, 27, 1087-1098.	0.3	51
26	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
27	Phenotypic patterns of desminopathy associated with three novel mutations in the desmin gene. Neuromuscular Disorders, 2007, 17, 443-450.	0.3	50
28	New insights into the protein aggregation pathology in myotilinopathy by combined proteomic and immunolocalization analyses. Acta Neuropathologica Communications, 2016, 4, 8.	2.4	50
29	Expression of mutant ubiquitin (UBB⁺¹) and p62 in myotilinopathies and desminopathies. Neuropathology and Applied Neurobiology, 2008, 34, 76-87.	1.8	48
30	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
31	Myofibrillar myopathies. Current Opinion in Neurology, 2013, 26, 527-535.	1.8	48
32	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. PLoS ONE, 2016, 11, e0156359.	1.1	48
33	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
34	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
35	Oxidative Stress in Desminopathies and Myotilinopathies: A Link between Oxidative Damage and Abnormal Protein Aggregation. Brain Pathology, 2007, 17, 377-388.	2.1	43
36	Prognostic value of X-chromosome inactivation in symptomatic female carriers of dystrophinopathy. Orphanet Journal of Rare Diseases, 2012, 7, 82.	1.2	40

#	Article	IF	Citations
37	Muscle imaging in muscle dystrophies produced by mutations in the EMD and LMNA genes. Neuromuscular Disorders, 2016, 26, 33-40.	0.3	40
38	Targeted Next-Generation Sequencing Reveals Novel TTN Mutations Causing Recessive Distal Titinopathy. Molecular Neurobiology, 2017, 54, 7212-7223.	1.9	38
39	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
40	Telethonin-deficiency initially presenting as a congenital muscular dystrophy. Neuromuscular Disorders, 2011, 21, 433-438.	0.3	37
41	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
42	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
43	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
44	Radiosensitive populations and recovery in X-ray-induced apoptosis in the developing cerebellum. Acta Neuropathologica, 1993, 86, 491-500.	3.9	35
45	Apoptosis is not the mechanism of cell death of muscle fibers in human muscular dystrophies and inflammatory myopathies. , 1997, 20, 1328-1330.		33
46	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
47	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
48	Bcl-2 and Bax protein expression in human myopathies. Journal of the Neurological Sciences, 1999, 164, 76-81.	0.3	32
49	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
50	Hereditary spastic paraparesis with dementia, amyotrophy and peripheral neuropathy. A neuropathological study. Neuropathology and Applied Neurobiology, 1995, 21, 255-261.	1.8	30
51	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
52	Impairment of protein degradation in myofibrillar myopathy caused by FLNC/filamin C mutations. Autophagy, 2013, 9, 422-423.	4.3	30
53	Hypoxia triggers IFN-I production in muscle: Implications in dermatomyositis. Scientific Reports, 2017, 7, 8595.	1.6	30
54	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

#	Article	IF	Citations
55	Expression of the intermediate filament protein synemin in myofibrillar myopathies and other muscle diseases. Acta Neuropathologica, 2003, 106, 1-7.	3.9	29
56	Different early pathogenesis in myotilinopathy compared to primary desminopathy. Neuromuscular Disorders, 2006, 16, 361-367.	0.3	29
57	Late-onset thymidine kinase 2 deficiency: a review of 18 cases. Orphanet Journal of Rare Diseases, 2019, 14, 100.	1.2	29
58	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
59	Mutations in TRIM63 cause an autosomal-recessive form of hypertrophic cardiomyopathy. Heart, 2020, 106, 1342-1348.	1.2	27
60	Targeted Next-Generation Sequencing in a Large Cohort of Genetically Undiagnosed Patients with Neuromuscular Disorders in Spain. Genes, 2020, 11, 539.	1.0	25
61	Heterozygous CAPN3 missense variants causing autosomalâ€dominant calpainopathy in seven unrelated families. Neuropathology and Applied Neurobiology, 2021, 47, 283-296.	1.8	23
62	Expression of myogenic regulatory factors (MRFs) in human neuromuscular disorders. Neuropathology and Applied Neurobiology, 1997, 23, 475-482.	1.8	22
63	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
64	Actininopathy: A new muscular dystrophy caused by ACTN2 dominant mutations. Annals of Neurology, 2019, 85, 899-906.	2.8	22
65	A novel thymidine phosphorylase mutation in a Spanish MNGIE patient. Journal of the Neurological Sciences, 2005, 228, 35-39.	0.3	20
66	Mutations in the J domain of DNAJB6 cause dominant distal myopathy. Neuromuscular Disorders, 2020, 30, 38-46.	0.3	20
67	Nemaline myopathy type 6: Clinical and myopathological features. Muscle and Nerve, 2010, 42, 901-907.	1.0	19
68	A Roma founder BIN1 mutation causes a novel phenotype of centronuclear myopathy with rigid spine. Neurology, 2018, 91, e339-e348.	1.5	18
69	CREB-1 and CREB-2 immunoreactivity in the rat brain. Brain Research, 1996, 712, 159-164.	1.1	16
70	A novel mutation in the caveolin-3 gene causing familial isolated hyperCKaemia. Neuromuscular Disorders, 2004, 14, 321-324.	0.3	16
71	Extralysosomal Protein Degradation in Myofibrillar Myopathies. Brain Pathology, 2009, 19, 507-515.	2.1	16
72	Novel mutation in TCAP manifesting with asymmetric calves and early-onset joint retractions. Neuromuscular Disorders, 2016, 26, 749-753.	0.3	16

#	Article	IF	Citations
73	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
74	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
75	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
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	Amoeboid microglial response following X-ray-induced apoptosis in the neonatal rat brain. Neuroscience Letters, 1995, 193, 109-112.	1.0	13
78	Overexpression of semicarbazide-sensitive amine oxidase in human myopathies. Muscle and Nerve, 2004, 29, 261-266.	1.0	13
79	Parvalbumin immunohistochemistry in denervated skeletal muscle. Neuropathology and Applied Neurobiology, 1994, 20, 495-500.	1.8	12
80	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
81	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
82	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	5.8	11
83	Diabetic muscular infarction. , 2000, 23, 825-826.		10
84	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
85	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
86	Cutaneous Neoplasms in Myotonic Dystrophy Type 1. Dermatology, 2016, 232, 700-703.	0.9	10
87	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
88	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
89	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
90	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

#	Article	IF	Citations
91	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
92	Muscle imaging in laminopathies: Synthesis study identifies meaningful muscles for followâ€up. Muscle and Nerve, 2018, 58, 812-817.	1.0	8
93	Adult-onset distal spinal muscular atrophy: a new phenotype associated with KIF5A mutations. Brain, 2019, 142, e66-e66.	3.7	8
94	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
95	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
96	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
97	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
98	Cylindrical spirals in two families: Clinical and genetic investigations. Neuromuscular Disorders, 2020, 30, 151-158.	0.3	7
99	Cell death induced by gamma irradiation of developing skeletal muscle. Journal of Anatomy, 1995, 187 () Tj ETQq1 1 0.784314 rgBT /Ov	0.9	7
100	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
101	246th ENMC International Workshop: Protein aggregate myopathies 24â€“26 May 2019, Hoofddorp, The Netherlands. Neuromuscular Disorders, 2021, 31, 158-166.	0.3	5
102	Postnatal development of parvalbumin immunoreactivity in striated muscles of the rat. Anatomy and Embryology, 1994, 190, 301-5.	1.5	4
103	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
104	Muscle MRI characteristic pattern for late-onset TK2 deficiency diagnosis. Journal of Neurology, 2022, 269, 3550-3562.	1.8	4
105	Dried Blood Spot for Screening for Late-Onset Pompe Disease: A Spanish Cohort. Journal of Neuromuscular Diseases, 2015, 2, S42-S42.	1.1	3
106	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
107	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
108	Expression of myogenic regulatory factors (MRFs) in human neuromuscular disorders. Neuropathology and Applied Neurobiology, 1997, 23, 475-482.	1.8	3

#	Article	IF	Citations
109	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
110	BNIP3 Is Involved in Muscle Fiber Atrophy in Late-Onset Pompe Disease Patients. American Journal of Pathology, 2022, , .	1.9	3
111	Congenital Muscular Dystrophy with Distinct CNS Involvement. Neuropediatrics, 1994, 25, 48-50.	0.3	2
112	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
113	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
114	SOD1 mutations in adultâ€onset distal spinal muscular atrophy. European Journal of Neurology, 2020, 27, e75-e76.	1.7	1
115	Myofibrillar Myopathies. , 2013, , 247-265.		1
116	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
117	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
118	G.P.38. Neuromuscular Disorders, 2014, 24, 806.	0.3	0
119	Pediatric onset of mitochondrial myopathy due to ANT1 mutation. Neuromuscular Disorders, 2016, 26, S176.	0.3	0
120	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
121	CONGENITAL MUSCULAR DYSTROPHIES. Neuromuscular Disorders, 2018, 28, S130.	0.3	0
122	Late onset distal myopathy: A new telethoninopathy. Neuromuscular Disorders, 2019, 29, 80-83.	0.3	0
123	High prevalence of paraspinal muscle involvement in adults with <scp>McArdle</scp> disease. Muscle and Nerve, 2022, , .	1.0	0