Olimpia Musumeci

List of Publications by Year
in descending order

141 papers	4,221 citations	101543 36 h-index	144013 57 g-index
143 all docs	143 docs citations	143 times ranked	7370 citing authors

#	Article	IF	Citations
1	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	7.6	385
2	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinsonâ€™s Disease Models. Cell Reports, 2018, 22, 2066-2079.	6.4	167
3	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
4	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	5.3	154
5	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
6	Observational clinical study in juvenile-adult glycogenosis type 2 patients undergoing enzyme replacement therapy for up to 4Âyears. Journal of Neurology, 2012, 259, 952-958.	3.6	117
7	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
8	Diagnosis of glycogenosis type II. Neurology, 2008, 71, S4-11.	1.1	87
9	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
10	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
11	MYH7-related myopathies: clinical, histopathological and imaging findings in a cohort of Italian patients. Orphanet Journal of Rare Diseases, 2016, 11, 91.	2.7	70
12	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
13	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. Neuromuscular Disorders, 2015, 25, 533-541.	0.6	65
14	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
15	Clinical and molecular aspects of 30 patients with late-onset Pompe disease (LOPD): unusual features and response to treatment. Journal of Neurology, 2015, 262, 968-978.	3.6	61
16	Safety and efficacy of avalglucosidase alfa versus alglucosidase alfa in patients with late-onset Pompe disease (COMET): a phase 3, randomised, multicentre trial. Lancet Neurology, The, 2021, 20, 1012-1026.	10.2	59
17	New motor outcome function measures in evaluation of Lateâ€Onset Pompe disease before and after enzyme replacement therapy. Muscle and Nerve, 2012, 45, 831-834.	2.2	56
18	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	3.6	55

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19	Late-onset Pompe disease (LOPD): Correlations between respiratory muscles CT and MRI features and pulmonary function. Molecular Genetics and Metabolism, 2013, 110, 290-296.	1.1	54
20	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
21	Management and treatment of glycogenosis type II. Neurology, 2008, 71, S12-36.	1.1	52
22	Multisystem late onset Pompe disease (LOPD): an update on clinical aspects. Annals of Translational Medicine, 2019, 7, 284-284.	1.7	52
23	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, jnnp-2014-310164.	1.9	50
24	Intragenic Inversion of mtDNA: A New Type of Pathogenic Mutation in a Patient with Mitochondrial Myopathy. American Journal of Human Genetics, 2000, 66, 1900-1904.	6.2	48
25	Italian validation of INQoL, a quality of life questionnaire for adults with muscle diseases. European Journal of Neurology, 2010, 17, 1178-1187.	3.3	48
26	Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE): Position paper on diagnosis, prognosis, and treatment by the <scp>MNGIE</scp> International Network. Journal of Inherited Metabolic Disease, 2021, 44, 376-387.	3.6	47
27	Clinical features and new molecular findings in Carnitine Palmitoyltransferase II (CPT II) deficiency. Journal of the Neurological Sciences, 2008, 266, 97-103.	0.6	46
28	Oxidative stress in myotonic dystrophy type 1. Free Radical Research, 2005, 39, 771-776.	3.3	45
29	Clinical features and new molecular findings in muscle phosphofructokinase deficiency (GSD type VII). Neuromuscular Disorders, 2012, 22, 325-330.	0.6	44
30	Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with CYP7B1 mutations. Clinical Genetics, 2012, 81, 150-157.	2.0	42
31	Safety and efficacy of cipaglucosidase alfa plus miglustat versus alglucosidase alfa plus placebo in late-onset Pompe disease (PROPEL): an international, randomised, double-blind, parallel-group, phase 3 trial. Lancet Neurology, The, 2021, 20, 1027-1037.	10.2	42
32	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	2.5	41
33	ZFYVE26/SPASTIZIN and SPG11/SPATACSIN mutations in hereditary spastic paraplegia types AR-SPG15 and AR-SPG11 have different effects on autophagy and endocytosis. Autophagy, 2019, 15, 34-57.	9.1	41
34	Muscle Phosphoglycerate Mutase Deficiency Revisited. Archives of Neurology, 2009, 66, 394-8.	4.5	40
35	Clinical and pathophysiological clues of respiratory dysfunction in late-onset Pompe disease: New insights from a comparative study by MRI and respiratory function assessment. Neuromuscular Disorders, 2015, 25, 852-858.	0.6	40
36	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.5	39

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37	Hepatic and neuromuscular forms of glycogenosis type III: nine mutations inAGL. Human Mutation, 2006, 27, 600-601.	2.5	38
38	Cerebellar ataxia and severe muscle <scp>CoQ₁₀</scp> deficiency in a patient with a novel mutation in <scp>ADCK3</scp>. Clinical Genetics, 2016, 90, 156-160.	2.0	38
39	â€œMitochondrial neuropathiesâ€: A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
40	Hippo signaling pathway is altered in Duchenne muscular dystrophy. PLoS ONE, 2018, 13, e0205514.	2.5	37
41	Central nervous system involvement in lateâ€onset Pompe disease: clues from neuroimaging and neuropsychological analysis. European Journal of Neurology, 2019, 26, 442.	3.3	35
42	Dichotomous metabolic networks govern human ILC2 proliferation and function. Nature Immunology, 2021, 22, 1367-1374.	14.5	34
43	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33
44	Intracranial arterial abnormalities in patients with late onset Pompe disease (LOPD). Journal of Inherited Metabolic Disease, 2016, 39, 391-398.	3.6	32
45	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32
46	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
47	Polarized mitochondria as guardians of NK cell fitness. Blood Advances, 2021, 5, 26-38.	5.2	32
48	Tarui disease and distal glycogenoses: clinical and genetic update. Acta Myologica, 2007, 26, 105-7.	1.5	32
49	Asymptomatic hyperCKemia in a case of Danon disease due to a missense mutation in Lamp-2 gene. Neuromuscular Disorders, 2005, 15, 409-411.	0.6	31
50	Cardiological manifestations of mitochondrial respiratory chain disorders. Acta Myologica, 2011, 30, 9-15.	1.5	31
51	Pathogenesis of the deafness-associated A1555G mitochondrial DNA mutation. Biochemical and Biophysical Research Communications, 2002, 293, 521-529.	2.1	30
52	Point mutations and a large intragenic deletion in SPG11 in complicated spastic paraplegia without thin corpus callosum. Journal of Medical Genetics, 2009, 46, 345-351.	3.2	30
53	Homozygosity for the common GAA gene splice site mutation c.-32-13T>G in Pompe disease is associated with the classical adult phenotypical spectrum. Neuromuscular Disorders, 2015, 25, 719-724.	0.6	29
54	Clinical, Molecular, and Functional Characterization of CLCN1 Mutations in Three Families with Recessive Myotonia Congenita. NeuroMolecular Medicine, 2015, 17, 285-296.	3.4	29

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55	Flecainide-Responsive Myotonia Permanens With SNEL Onset: A New Case and Literature Review. Pediatrics, 2016, 137, .	2.1	27
56	Apoptosis in metabolic myopathies. NeuroReport, 1998, 9, 2431-2435.	1.2	26
57	Auditory system involvement in late onset Pompe disease: A study of 20 Italian patients. Molecular Genetics and Metabolism, 2012, 107, 480-484.	1.1	26
58	A Novel Mutation in the Mitochondrial DNA Transfer Ribonucleic AcidAsp Gene in a Child With Myoclonic Epilepsy and Psychomotor Regression. Journal of Child Neurology, 1999, 14, 610-613.	1.4	25
59	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.6	25
60	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
61	Estimating the impact of COVID-19 pandemic on services provided by Italian Neuromuscular Centers: an Italian Association of Myology survey of the acute phase. Acta Myologica, 2020, 39, 57-66.	1.5	24
62	Effects of shortâ€toâ€long term enzyme replacement therapy (ERT) on skeletal muscle tissue in late onset Pompe disease (LOPD). Neuropathology and Applied Neurobiology, 2018, 44, 449-462.	3.2	23
63	Data from the European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC). Orphanet Journal of Rare Diseases, 2020, 15, 330.	2.7	23
64	Recurrent rhabdomyolysis due to muscle Î²-enolase deficiency: very rare or underestimated?. Journal of Neurology, 2014, 261, 2424-2428.	3.6	22
65	Risk of Myopathy in Patients in Therapy with Statins: Identification of Biological Markers in a Pilot Study. Frontiers in Pharmacology, 2017, 8, 500.	3.5	22
66	microRNAs as biomarkers in Pompe disease. Genetics in Medicine, 2019, 21, 591-600.	2.4	22
67	A novel mutation in KIF5A gene causing hereditary spastic paraplegia with axonal neuropathy. Neurological Sciences, 2011, 32, 665-668.	1.9	21
68	Role of the cardio-pulmonary exercise test and six-minute walking test in the evaluation of exercise performance in patients with late-onset Pompe disease. Neuromuscular Disorders, 2015, 25, 542-547.	0.6	21
69	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
70	Update on diagnostics of metabolic myopathies. Current Opinion in Neurology, 2017, 30, 553-562.	3.6	20
71	Genetic neuromuscular disorders: living the era of a therapeutic revolution. Part 2: diseases of motor neuron and skeletal muscle. Neurological Sciences, 2019, 40, 671-681.	1.9	20
72	Early is better? A new algorithm for early diagnosis in late onset Pompe disease (LOPD). Acta Myologica, 2013, 32, 78-81.	1.5	20

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73	A Novel Missense Mutation (W797R) in the Myophosphorylase Gene in Spanish Patients With McArdle Disease. Archives of Neurology, 2000, 57, 217.	4.5	19
74	Sporadic late-onset nemaline myopathy in a woman with multiple myeloma successfully treated with lenalidomide/dexamethasone. Muscle and Nerve, 2015, 51, 934-935.	2.2	19
75	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
76	Posterior reversible encephalopathy syndrome (PRES) and infection: a systematic review of the literature. Neurological Sciences, 2019, 40, 915-922.	1.9	19
77	Evidence of cardiovascular autonomic impairment in mitochondrial disorders. Journal of Neurology, 2007, 254, 1498-1503.	3.6	17
78	Ultrasound assessment of diaphragm function in patients with late-onset Pompe disease. Neurological Sciences, 2020, 41, 2175-2184.	1.9	17
79	Statin-Induced Myopathy: Translational Studies from Preclinical to Clinical Evidence. International Journal of Molecular Sciences, 2021, 22, 2070.	4.1	17
80	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	3.6	15
81	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.6	15
82	Favourable course in a cohort of Parkinsonâ€™s disease patients infected by SARS-CoV-2: a single-centre experience. Neurological Sciences, 2021, 42, 811-816.	1.9	15
83	Diseases of Oxidative Phosphorylation Due to mtDNA Mutations. Seminars in Neurology, 2001, 21, 251-260.	1.4	14
84	Opposed-phase MR imaging of lipid storage myopathy in a case of Chanarinâ€“Dorfman disease. Skeletal Radiology, 2008, 37, 1053-1057.	2.0	14
85	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	1.4	14
86	Molecular Genetics Overview of Primary Mitochondrial Myopathies. Journal of Clinical Medicine, 2022, 11, 632.	2.4	14
87	Diagnostic tools in late onset Pompe disease (LOPD). Annals of Translational Medicine, 2019, 7, 286-286.	1.7	13
88	Amyloid myopathy presenting with rhabdomyolysis: Evidence of complement activation. Neuromuscular Disorders, 2006, 16, 514-517.	0.6	12
89	Myasthenia Gravis: Unusual Presentations and Diagnostic Pitfalls. Journal of Neuromuscular Diseases, 2016, 3, 413-418.	2.6	12
90	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12

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91	Molecular genetic study of myophosphorylase deficiency (McArdle's disease) in two Yemenite-Jewish families. Neuromuscular Disorders, 2002, 12, 824-827.	0.6	10
92	Immunohistochemical analysis of human skeletal muscle AMP deaminase deficiency. Evidence of a correlation between the muscle HPRG content and the level of the residual AMP deaminase activity. Journal of Muscle Research and Cell Motility, 2006, 27, 83-92.	2.0	10
93	Ischemic stroke due to hypoperfusion in a patient with a previously unrecognized Danon disease. Neuromuscular Disorders, 2016, 26, 890-894.	0.6	10
94	Primary mitochondrial myopathy. Neurology: Genetics, 2020, 6, e519.	1.9	10
95	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. Journal of Neurology, 2022, 269, 1413-1421.	3.6	10
96	Identification of the infant-type R631C mutation in patients with the benign muscular form of CPT2 deficiency. Neuromuscular Disorders, 2007, 17, 960-963.	0.6	9
97	Muscle pain in mitochondrial diseases: a picture from the Italian network. Journal of Neurology, 2019, 266, 953-959.	3.6	9
98	MRI findings of neutrophilic fasciitis in a patient with acute febrile neutrophilic dermatosis (Sweetâ€™s) Tj ETQq0 0 0 rgBT /Overlock 10	2.0	8
99	Acute parkinsonism as first manifestation of systemic lupus erythematosus unmasked by CMV infection. Neurological Sciences, 2014, 35, 2019-2021.	1.9	8
100	Assessing the Role of Anti rh-GAA in Modulating Response to ERT in a Late-Onset Pompe Disease Cohort from the Italian GSDII Study Group. Advances in Therapy, 2019, 36, 1177-1189.	2.9	8
101	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. Journal of Clinical Medicine, 2021, 10, 2063.	2.4	8
102	A new stop codon mutation (Y52X) in the myophosphorylase gene in a Greek patient with McArdle's disease. Journal of the Neurological Sciences, 2002, 194, 83-86.	0.6	7
103	Vacuolated PAS-Positive Lymphocytes on Blood Smear: An Easy Screening Tool and a Possible Biomarker for Monitoring Therapeutic Responses in Late Onset Pompe Disease (LOPD). Frontiers in Neurology, 2018, 9, 880.	2.4	7
104	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
105	Diagnostic Challenges in Late Onset Multiple Acyl-CoA Dehydrogenase Deficiency: Clinical, Morphological, and Genetic Aspects. Frontiers in Neurology, 2022, 13, 815523.	2.4	7
106	Identification of a novel mutation in Cu/Zn superoxide dismutase gene associated with familial amyotrophic lateral sclerosis. Journal of the Neurological Sciences, 2002, 198, 17-19.	0.6	6
107	MicroRNA signatures predict dysregulated vitamin D receptor and calcium pathways status in limb girdle muscle dystrophies (LGMD) 2A/2B. Cell Biochemistry and Function, 2016, 34, 414-422.	2.9	5
108	Novel SHOX Gene Mutation in a Short Boy with Becker Muscular Dystrophy: Double Trouble in Two Adjacent Genes. Hormone Research in Paediatrics, 2008, 69, 124-128.	1.8	4

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109	Stiffness as a presenting symptom of an odd clinical condition caused by multiple sclerosis and myotonia congenita. Neuromuscular Disorders, 2013, 23, 52-55.	0.6	4
110	Methotrexate as a Steroid-Sparing Agent in Myasthenia Gravis: A Preliminary Retrospective Study. Journal of Clinical Neuromuscular Disease, 2021, 23, 61-65.	0.7	4
111	Intracranial aneurysm management in patients with late-onset Pompe disease (LOPD). Neurological Sciences, 2021, 42, 2411-2419.	1.9	3
112	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. Mitochondrion, 2021, 58, 243-245.	3.4	3
113	Spectrum of movement disorders in mitochondrial diseases. , 0, , .		3
114	Mitochondrial Disorders in Adults. Current Molecular Medicine, 2014, 14, 1001-1008.	1.3	3
115	T.P.34. Neuromuscular Disorders, 2014, 24, 874-875.	0.6	2
116	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
117	Mitochondrial Disease (MELAS Syndrome) Discovered at the Start of Pregnancy in a Patient with Advanced CKD: A Clinical and Ethical Challenge. Journal of Clinical Medicine, 2019, 8, 303.	2.4	2
118	Late and Severe Myopathy in a Patient With Glycogenosis VII Worsened by Cyclosporine and Amiodarone. Frontiers in Neurology, 2019, 10, 77.	2.4	2
119	A Family With a Complex Phenotype Caused by Two Different Rare Metabolic Disorders: GLUT1 and Very-Long-Chain Fatty Acid Dehydrogenase (VLCAD) Deficiencies. Frontiers in Neurology, 2020, 11, 514.	2.4	2
120	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	1.9	2
121	Breathing Pattern and Central Ventilatory Drive in Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S49.	2.6	2
122	G.P.245. Neuromuscular Disorders, 2014, 24, 890.	0.6	1
123	Energy metabolism during exercise in patients with Î²â€enolase deficiency (GSDXIII). JIMD Reports, 2021, 61, 60-66.	1.5	1
124	Intracranial Arterial Abnormalities in Patients with Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S48.	2.6	1
125	A new phenotype of muscle glycogen synthase deficiency (GSD0B) characterized by an adult onset myopathy without cardiomyopathy. Neuromuscular Disorders, 2022, 32, 582-589.	0.6	1
126	Atypical rat cerebellar immunoreactivity in a patient with familial amyloid polyneuropathy. Journal of the Peripheral Nervous System, 2004, 9, 121-122.	3.1	0

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127	G.P.1 08 Phenotypeâ€“genotype correlation in two families with muscle phosphofructokinase deficiency. Neuromuscular Disorders, 2006, 16, 656.	0.6	0
128	G.P.7 02 Amyloid myopathy presenting with rhabdomyolysis: Evidence of complement activation. Neuromuscular Disorders, 2006, 16, 701-702.	0.6	0
129	G.P.3.09 MicroRNA expression in Duchenne and Becker muscular dystrophy. Neuromuscular Disorders, 2007, 17, 784-785.	0.6	0
130	G.P.11.05 A life threatening case of Î±-enolase deficiency. Neuromuscular Disorders, 2009, 19, 624-625.	0.6	0
131	Treatment Guidelines for Pompe Disease: A South European Perspective. Clinical Therapeutics, 2011, 33, S8.	2.5	0
132	G.O.7. Neuromuscular Disorders, 2014, 24, 851.	0.6	0
133	T.P.16. Neuromuscular Disorders, 2014, 24, 869.	0.6	0
134	8 Years of Experience with Alglucosidase Alpha Treatment: Facts and Perspectives. Journal of Neuromuscular Diseases, 2015, 2, S4-S4.	2.6	0
135	Clinical and molecular features of a large cohort of Italian McArdle patients. Neuromuscular Disorders, 2015, 25, S219.	0.6	0
136	The EUROMAC registry for rare glycogen storage diseases: preliminary report. Neuromuscular Disorders, 2017, 27, S203-S204.	0.6	0
137	POMPE DISEASE. Neuromuscular Disorders, 2021, 31, S109.	0.6	0
138	Adult onset cerebellar ataxia due to novel mutations in BRAT1. Journal of the Neurological Sciences, 2021, 429, 118261.	0.6	0
139	Extension to the heart of metastatic lung cancer presenting as acute neurological syndrome: The key role of echocardiography. Journal of Cardiovascular Echography, 2014, 24, 89.	0.4	0
140	Right ventricular obstructive hypertrophic cardiomyopathy in primary myo-adenylate deaminase deficiency. Acta Myologica, 2011, 30, 46-8.	1.5	0
141	Case Report: Thymidine Kinase 2 (TK2) Deficiency: A Novel Mutation Associated With Childhood-Onset Mitochondrial Myopathy and Atypical Progression. Frontiers in Neurology, 2022, 13, 857279.	2.4	0