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	Adult-onset mitochondrial movement disorders: a national picture from the Italian Network. Journal of Neurology, 2022, 269, 1413-1421.	3.6	10
2	Molecular Genetics Overview of Primary Mitochondrial Myopathies. Journal of Clinical Medicine, 2022, 11, 632.	2.4	14
3	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
4	Diagnostic Challenges in Late Onset Multiple Acyl-CoA Dehydrogenase Deficiency: Clinical, Morphological, and Genetic Aspects. Frontiers in Neurology, 2022, 13, 815523.	2.4	7
5	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
6	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
7	Intracranial aneurysm management in patients with late-onset Pompe disease (LOPD). Neurological Sciences, 2021, 42, 2411-2419.	1.9	3
8	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
9	Statin-Induced Myopathy: Translational Studies from Preclinical to Clinical Evidence. International Journal of Molecular Sciences, 2021, 22, 2070.	4.1	17
10	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
11	SARS-CoV-2 infection in patients with primary mitochondrial diseases: Features and outcomes in Italy. Mitochondrion, 2021, 58, 243-245.	3.4	3
12	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
13	Energy metabolism during exercise in patients with Î²â€enolase deficiency (GSDXIII). JIMD Reports, 2021, 61, 60-66.	1.5	1
14	NGS in Hereditary Ataxia: When Rare Becomes Frequent. International Journal of Molecular Sciences, 2021, 22, 8490.	4.1	12
15	POMPE DISEASE. Neuromuscular Disorders, 2021, 31, S109.	0.6	0
16	Polarized mitochondria as guardians of NK cell fitness. Blood Advances, 2021, 5, 26-38.	5.2	32
17	Adult onset cerebellar ataxia due to novel mutations in BRAT1. Journal of the Neurological Sciences, 2021, 429, 118261.	0.6	0
18	Dichotomous metabolic networks govern human ILC2 proliferation and function. Nature Immunology, 2021, 22, 1367-1374.	14.5	34

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19	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
20	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
21	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
22	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	1.4	14
23	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
24	Primary mitochondrial myopathy. Neurology: Genetics, 2020, 6, e519.	1.9	10
25	Ultrasound assessment of diaphragm function in patients with late-onset Pompe disease. Neurological Sciences, 2020, 41, 2175-2184.	1.9	17
26	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
27	Awareness of rare and genetic neurological diseases among italian neurologist. A national survey. Neurological Sciences, 2020, 41, 1567-1570.	1.9	2
28	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
29	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
30	microRNAs as biomarkers in Pompe disease. Genetics in Medicine, 2019, 21, 591-600.	2.4	22
31	Muscle pain in mitochondrial diseases: a picture from the Italian network. Journal of Neurology, 2019, 266, 953-959.	3.6	9
32	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
33	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
34	Lipomatosis Incidence and Characteristics in an Italian Cohort of Mitochondrial Patients. Frontiers in Neurology, 2019, 10, 160.	2.4	19
35	A genetic modifier of symptom onset in Pompe disease. EBioMedicine, 2019, 43, 553-561.	6.1	32
36	Late and Severe Myopathy in a Patient With Glycogenosis VII Worsened by Cyclosporine and Amiodarone. Frontiers in Neurology, 2019, 10, 77.	2.4	2

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37	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
38	Posterior reversible encephalopathy syndrome (PRES) and infection: a systematic review of the literature. Neurological Sciences, 2019, 40, 915-922.	1.9	19
39	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
40	Diagnostic tools in late onset Pompe disease (LOPD). Annals of Translational Medicine, 2019, 7, 286-286.	1.7	13
41	Multisystem late onset Pompe disease (LOPD): an update on clinical aspects. Annals of Translational Medicine, 2019, 7, 284-284.	1.7	52
42	Pharmacological Inhibition of Necroptosis Protects from Dopaminergic Neuronal Cell Death in Parkinsonâ€™s Disease Models. Cell Reports, 2018, 22, 2066-2079.	6.4	167
43	A mobile app for patients with Pompe disease and its possible clinical applications. Neuromuscular Disorders, 2018, 28, 471-475.	0.6	15
44	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. Neuromuscular Disorders, 2018, 28, 586-591.	0.6	24
45	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
46	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
47	Next Generation Molecular Diagnosis of Hereditary Spastic Paraplegias: An Italian Cross-Sectional Study. Frontiers in Neurology, 2018, 9, 981.	2.4	64
48	Hippo signaling pathway is altered in Duchenne muscular dystrophy. PLoS ONE, 2018, 13, e0205514.	2.5	37
49	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. Genes, 2018, 9, 524.	2.4	7
50	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. Muscle and Nerve, 2017, 55, 55-68.	2.2	86
51	Muscle MRI in neutral lipid storage disease (NLSD). Journal of Neurology, 2017, 264, 1334-1342.	3.6	15
52	Update on diagnostics of metabolic myopathies. Current Opinion in Neurology, 2017, 30, 553-562.	3.6	20
53	The EUROMAC registry for rare glycogen storage diseases: preliminary report. Neuromuscular Disorders, 2017, 27, S203-S204.	0.6	0
54	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. Journal of Neurology, 2017, 264, 1777-1784.	3.6	32

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55	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
56	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
57	Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. PLoS ONE, 2016, 11, e0153283.	2.5	41
58	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
59	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
60	Flecainide-Responsive Myotonia Permanens With SNEL Onset: A New Case and Literature Review. Pediatrics, 2016, 137, .	2.1	27
61	Ischemic stroke due to hypoperfusion in a patient with a previously unrecognized Danon disease. Neuromuscular Disorders, 2016, 26, 890-894.	0.6	10
62	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
63	Myasthenia Gravis: Unusual Presentations and Diagnostic Pitfalls. Journal of Neuromuscular Diseases, 2016, 3, 413-418.	2.6	12
64	The genetic basis of undiagnosed muscular dystrophies and myopathies. Neurology, 2016, 87, 71-76.	1.1	92
65	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. Neuromuscular Disorders, 2016, 26, 292-299.	0.6	25
66	â€œMitochondrial neuropathiesâ€• A survey from the large cohort of the Italian Network. Neuromuscular Disorders, 2016, 26, 272-276.	0.6	37
67	Intracranial arterial abnormalities in patients with late onset Pompe disease (LOPD). Journal of Inherited Metabolic Disease, 2016, 39, 391-398.	3.6	32
68	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
69	8 Years of Experience with Alglucosidase Alpha Treatment: Facts and Perspectives. Journal of Neuromuscular Diseases, 2015, 2, S4-S4.	2.6	0
70	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
71	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
72	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

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73	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. Neuromuscular Disorders, 2015, 25, 533-541.	0.6	65
74	Syndromic parkinsonism and dementia associated with <scp>OPA</scp>1 missense mutations. Annals of Neurology, 2015, 78, 21-38.	5.3	154
75	Redefining phenotypes associated with mitochondrial DNA single deletion. Journal of Neurology, 2015, 262, 1301-1309.	3.6	68
76	Variants in KIF1A gene in dominant and sporadic forms of hereditary spastic paraparesis. Journal of Neurology, 2015, 262, 2684-2690.	3.6	55
77	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
78	Clinical, Molecular, and Functional Characterization of CLCN1 Mutations in Three Families with Recessive Myotonia Congenita. NeuroMolecular Medicine, 2015, 17, 285-296.	3.4	29
79	Clinical and molecular features of a large cohort of Italian McArdle patients. Neuromuscular Disorders, 2015, 25, S219.	0.6	0
80	Intracranial Arterial Abnormalities in Patients with Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S48.	2.6	1
81	Breathing Pattern and Central Ventilatory Drive in Late-Onset Pompe Disease. Journal of Neuromuscular Diseases, 2015, 2, S49.	2.6	2
82	Recurrent rhabdomyolysis due to muscle Î²-enolase deficiency: very rare or underestimated?. Journal of Neurology, 2014, 261, 2424-2428.	3.6	22
83	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. Journal of Neurology, 2014, 261, 504-510.	3.6	119
84	T.P.34. Neuromuscular Disorders, 2014, 24, 874-875.	0.6	2
85	G.P.251. Neuromuscular Disorders, 2014, 24, 892.	0.6	2
86	G.O.7. Neuromuscular Disorders, 2014, 24, 851.	0.6	0
87	T.P.16. Neuromuscular Disorders, 2014, 24, 869.	0.6	0
88	G.P.245. Neuromuscular Disorders, 2014, 24, 890.	0.6	1
89	Acute parkinsonism as first manifestation of systemic lupus erythematosus unmasked by CMV infection. Neurological Sciences, 2014, 35, 2019-2021.	1.9	8
90	Myoclonus in mitochondrial disorders. Movement Disorders, 2014, 29, 722-728.	3.9	33

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91	Mitochondrial Disorders in Adults. Current Molecular Medicine, 2014, 14, 1001-1008.	1.3	3
92	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
93	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
94	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
95	Phenotypic heterogeneity of the 8344A>G mtDNA â€œMERRFâ€•mutation. Neurology, 2013, 80, 2049-2054.	1.1	157
96	Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. Brain, 2013, 136, 3119-3139.	7.6	74
97	Early is better? A new algorithm for early diagnosis in late onset Pompe disease (LOPD). Acta Myologica, 2013, 32, 78-81.	1.5	20
98	Clinical features and new molecular findings in muscle phosphofructokinase deficiency (GSD type VII). Neuromuscular Disorders, 2012, 22, 325-330.	0.6	44
99	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
100	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
101	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
102	Clinical phenotype variability in patients with hereditary spastic paraplegia type 5 associated with CYP7B1 mutations. Clinical Genetics, 2012, 81, 150-157.	2.0	42
103	Treatment Guidelines for Pompe Disease: A South European Perspective. Clinical Therapeutics, 2011, 33, S8.	2.5	0
104	A novel mutation in KIF5A gene causing hereditary spastic paraplegia with axonal neuropathy. Neurological Sciences, 2011, 32, 665-668.	1.9	21
105	MRI findings of neutrophilic fasciitis in a patient with acute febrile neutrophilic dermatosis (Sweetâ€™s) Tj ETQq1 1 0.784314 rgBT /O	2.0	8
106	Right ventricular obstructive hypertrophic cardiomyopathy in primary myo-adenylate deaminase deficiency. Acta Myologica, 2011, 30, 46-8.	1.5	0
107	Cardiological manifestations of mitochondrial respiratory chain disorders. Acta Myologica, 2011, 30, 9-15.	1.5	31
108	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

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109	Multi-system neurological disease is common in patients with OPA1 mutations. Brain, 2010, 133, 771-786.	7.6	385
110	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
111	Muscle Phosphoglycerate Mutase Deficiency Revisited. Archives of Neurology, 2009, 66, 394-8.	4.5	40
112	G.P.11.05 A life threatening case of Î±-enolase deficiency. Neuromuscular Disorders, 2009, 19, 624-625.	0.6	0
113	Opposed-phase MR imaging of lipid storage myopathy in a case of Chanarinâ€“Dorfman disease. Skeletal Radiology, 2008, 37, 1053-1057.	2.0	14
114	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
115	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
116	Management and treatment of glycogenosis type II. Neurology, 2008, 71, S12-36.	1.1	52
117	Diagnosis of glycogenosis type II. Neurology, 2008, 71, S4-11.	1.1	87
118	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
119	G.P.3.09 MicroRNA expression in Duchenne and Becker muscular dystrophy. Neuromuscular Disorders, 2007, 17, 784-785.	0.6	0
120	Evidence of cardiovascular autonomic impairment in mitochondrial disorders. Journal of Neurology, 2007, 254, 1498-1503.	3.6	17
121	Tarui disease and distal glycogenoses: clinical and genetic update. Acta Myologica, 2007, 26, 105-7.	1.5	32
122	G.P.1 08 Phenotypeâ€“genotype correlation in two families with muscle phosphofructokinase deficiency. Neuromuscular Disorders, 2006, 16, 656.	0.6	0
123	G.P.7 02 Amyloid myopathy presenting with rhabdomyolysis: Evidence of complement activation. Neuromuscular Disorders, 2006, 16, 701-702.	0.6	0
124	Amyloid myopathy presenting with rhabdomyolysis: Evidence of complement activation. Neuromuscular Disorders, 2006, 16, 514-517.	0.6	12
125	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
126	Hepatic and neuromuscular forms of glycogenosis type III: nine mutations inAGL. Human Mutation, 2006, 27, 600-601.	2.5	38

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127	McArdle disease: the mutation spectrum ofPYGMin a large Italian cohort. Human Mutation, 2006, 27, 718-718.	2.5	52
128	Eight Novel Mutations in SPG4 in a Large Sample of Patients With Hereditary Spastic Paraplegia. Archives of Neurology, 2006, 63, 750.	4.5	39
129	Oxidative stress in myotonic dystrophy type 1. Free Radical Research, 2005, 39, 771-776.	3.3	45
130	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
131	Atypical rat cerebellar immunoreactivity in a patient with familial amyloid polyneuropathy. Journal of the Peripheral Nervous System, 2004, 9, 121-122.	3.1	0
132	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
133	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
134	Pathogenesis of the deafness-associated A1555G mitochondrial DNA mutation. Biochemical and Biophysical Research Communications, 2002, 293, 521-529.	2.1	30
135	Molecular genetic study of myophosphorylase deficiency (McArdle's disease) in two Yemenite-Jewish families. Neuromuscular Disorders, 2002, 12, 824-827.	0.6	10
136	Diseases of Oxidative Phosphorylation Due to mtDNA Mutations. Seminars in Neurology, 2001, 21, 251-260.	1.4	14
137	A Novel Missense Mutation (W797R) in the Myophosphorylase Gene in Spanish Patients With McArdle Disease. Archives of Neurology, 2000, 57, 217.	4.5	19
138	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
139	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
140	Apoptosis in metabolic myopathies. NeuroReport, 1998, 9, 2431-2435.	1.2	26
141	Spectrum of movement disorders in mitochondrial diseases. , 0, , .		3