Alessandro Prelle

List of Publications by Year
in descending order

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69 papers	4,326 citations	126858 33 h-index	106281 65 g-index
71 all docs	71 docs citations	71 times ranked	5824 citing authors

#	Article	IF	Citations
1	Loss of ETHE1, a mitochondrial dioxygenase, causes fatal sulfide toxicity in ethylmalonic encephalopathy. Nature Medicine, 2009, 15, 200-205.	15.2	358
2	Increased longevity and refractoriness to Ca2+-dependent neurodegeneration in Surf1 knockout mice. Human Molecular Genetics, 2007, 16, 431-444.	1.4	279
3	Cytochromec Oxidase subunit I microdeletion in a patient with motor neuron disease. Annals of Neurology, 1998, 43, 110-116.	2.8	251
4	Facioscapulohumeral muscular dystrophy in mice overexpressing FRG1. Nature, 2006, 439, 973-977.	13.7	200
5	Early vacuolization and mitochondrial damage in motor neurons of FALS mice are not associated with apoptosis or with changes in cytochrome oxidase histochemical reactivity. Journal of the Neurological Sciences, 2001, 191, 25-33.	0.3	185
6	Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72 -associated amyotrophic lateral sclerosis. Science Translational Medicine, 2017, 9, .	5.8	179
7	Clinical, molecular, and protein correlations in a large sample of genetically diagnosed Italian limb girdle muscular dystrophy patients. Human Mutation, 2008, 29, 258-266.	1.1	162
8	Muscle Coenzyme Q10 Level in Statin-Related Myopathy. Archives of Neurology, 2005, 62, 1709.	4.9	158
9	Cognitive impairment in Duchenne muscular dystrophy. Neuromuscular Disorders, 1994, 4, 359-369.	0.3	152
10	Divergent brain network connectivity in amyotrophic lateral sclerosis. Neurobiology of Aging, 2013, 34, 419-427.	1.5	133
11	?-enolase deficiency, a new metabolic myopathy of distal glycolysis. Annals of Neurology, 2001, 50, 202-207.	2.8	125
12	Mitochondrial Respiratory Chain Dysfunction in Muscle From Patients With Amyotrophic Lateral Sclerosis. Archives of Neurology, 2010, 67, 849-54.	4.9	122
13	Genotype to phenotype correlations in mitochondrial encephalomyopathies associated with the A3243G mutation of mitochondrial DNA. Journal of Neurology, 1995, 242, 304-312.	1.8	115
14	The Cortical Signature of Amyotrophic Lateral Sclerosis. PLoS ONE, 2012, 7, e42816.	1.1	108
15	Retrospective study of a large population of patients with asymptomatic or minimally symptomatic raised serum creatine kinase levels. Journal of Neurology, 2002, 249, 305-311.	1.8	100
16	Progressive cytochromec oxidase deficiency in a case of earns-sayre syndrome: Morphological, immunological, and biochemical studies in muscle biopsies and autopsy tissues. Annals of Neurology, 1987, 21, 564-572.	2.8	96
17	TARDBP (TDPâ€43) sequence analysis in patients with familial and sporadic ALS: identification of two novel mutations. European Journal of Neurology, 2009, 16, 727-732.	1.7	93
18	Decorin and biglycan expression is differentially altered in several muscular dystrophies. Brain, 2005, 128, 2546-2555.	3.7	87

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19	Chronic Exposure to Sulfide Causes Accelerated Degradation of Cytochrome c Oxidase in Ethylmalonic Encephalopathy. Antioxidants and Redox Signaling, 2011, 15, 353-362.	2.5	80
20	Constitutive knockout of Surf1 is associated with high embryonic lethality, mitochondrial disease and cytochrome c oxidase deficiency in mice. Human Molecular Genetics, 2003, 12, 399-413.	1.4	74
21	A Subpopulation of Murine Bone Marrow Cells Fully Differentiates along the Myogenic Pathway and Participates in Muscle Repair in the mdx Dystrophic Mouse. Experimental Cell Research, 2002, 277, 74-85.	1.2	70
22	New Mutations in TK2 Gene Associated With Mitochondrial DNA Depletion. Pediatric Neurology, 2006, 34, 177-185.	1.0	63
23	Early-onset encephalomyopathy associated with tissue-specific mitochondrial DNA depletion: A morphological, biochemical and molecular-genetic study. Journal of Neurology, 1995, 242, 547-556.	1.8	60
24	Brain Activation in Normal Subjects and in Patients Affected by Mitochondrial Disease without Clinical Central Nervous System Involvement: A Phosphorus Magnetic Resonance Spectroscopy Study. Journal of Cerebral Blood Flow and Metabolism, 2001, 21, 85-91.	2.4	57
25	MRI predictors of longâ€term evolution in amyotrophic lateral sclerosis. European Journal of Neuroscience, 2010, 32, 1490-1496.	1.2	53
26	Retrospective study of a large population of patients affected with mitochondrial disorders: clinical, morphological and molecular genetic evaluation. Journal of Neurology, 2001, 248, 778-788.	1.8	45
27	Molecular analysis of LGMD-2B and MM patients: identification of novel DYSF mutations and possible founder effect in the Italian population. Neuromuscular Disorders, 2003, 13, 788-795.	0.3	45
28	Impaired expression of insulinâ€like growth factorâ€1 system in skeletal muscle of amyotrophic lateral sclerosis patients. Muscle and Nerve, 2012, 45, 200-208.	1.0	43
29	Absence of angiogenic genes modification in Italian ALS patients. Neurobiology of Aging, 2008, 29, 314-316.	1.5	41
30	Multiple deletions of mitochondrial DNA in sporadic and atypical cases of encephalomyopathy. Journal of the Neurological Sciences, 1994, 123, 74-79.	0.3	40
31	High mutational burden in the mtDNA control region from aged muscles: a single-fiber study. Neurobiology of Aging, 2003, 24, 829-838.	1.5	40
32	Congenital myopathy associated with abnormal accumulation of desmin and dystrophin. Neuromuscular Disorders, 1992, 2, 169-175.	0.3	38
33	OXPHOS defects and mitochondrial DNA mutations in cardiomyopathy. Muscle and Nerve, 1995, 18, S170-S174.	1.0	33
34	Primary beta-sarcoglycanopathy manifesting as recurrent exercise-induced myoglobinuria. Neuromuscular Disorders, 2001, 11, 389-394.	0.3	33
35	Novel missense mutation and large deletion ofGNE gene in autosomal-recessive inclusion-body myopathy. Muscle and Nerve, 2003, 28, 113-117.	1.0	32
36	Familial mtDNA T8993C transition causing both the NARP and the MILS phenotype in the same generation. Journal of Neurology, 2003, 250, 1498-1500.	1.8	28

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37	A novel mitochondrial tRNA Ile point mutation in chronic progressive external ophthalmoplegia. Journal of Neurology, 1998, 245, 755-758.	1.8	27
38	Cytochrome Oxidase Deficiency: Clinical and Biochemical Heterogeneity. Annals of the New York Academy of Sciences, 1986, 488, 19-32.	1.8	26
39	A G+1->A transversion at the 5' splice site of intron 69 of the dystrophin gene causing the absence of peripheral nerve Dp 116 and severe clinical involvement in a DMD patient. Human Molecular Genetics, 1995, 4, 2171-2174.	1.4	26
40	Sarcoglycan deficiency in a large Italian population of myopathic patients. Acta Neuropathologica, 1998, 96, 509-514.	3.9	25
41	Synergistic effect of Â-amyloid protein and interferon gamma on nitric oxide production by C2C12 muscle cells. Brain, 2000, 123, 374-379.	3.7	24
42	New molecular findings in congenital myopathies due to selenoprotein N gene mutations. Journal of the Neurological Sciences, 2011, 300, 107-113.	0.3	23
43	Chronic progressive external ophthalmoplegia: A correlative study of quantitative molecular data and histochemical and biochemical profile. Journal of the Neurological Sciences, 1994, 123, 140-146.	0.3	22
44	Asymptomatic familial hyperCKemia associated with desmin accumulation in skeletal muscle. Journal of the Neurological Sciences, 1996, 140, 132-136.	0.3	22
45	Follow-up of a large population of asymptomatic/oligosymptomatic hyperckemic subjects. Journal of Neurology, 2006, 253, 1399-1403.	1.8	22
46	An intragenic deletion/inversion event in the DMD gene determines a novel exon creation and results in a BMD phenotype. Human Genetics, 2004, 115, 13-18.	1.8	19
47	Multiple sclerosis and mitochondrial myopathy: An unusual combination of diseases. Journal of Neurology, 1994, 241, 511-516.	1.8	18
48	Steroid-responsive Hashimoto encephalopathy mimicking Creutzfeldtâ€“Jakob disease. Neurological Sciences, 2011, 32, 719-722.	0.9	16
49	A case of CPT deficiency, homoplasmic mtDNA mutation and ragged red fibers at muscle biopsy. Journal of the Neurological Sciences, 2005, 239, 21-24.	0.3	15
50	Appearance and localization of dystrophin in normal human fetal muscle. International Journal of Developmental Neuroscience, 1991, 9, 607-612.	0.7	14
51	Multiple deletions of mitochondrial DNA in a patient with periodic attacks of paralysis. Journal of the Neurological Sciences, 1993, 117, 24-27.	0.3	14
52	Adult Polyglucosan Body Disease: Clinical and histological heterogeneity of a large Italian family. Neuromuscular Disorders, 2015, 25, 423-428.	0.3	14
53	Utrophin expression during human fetal development. International Journal of Developmental Neuroscience, 1995, 13, 585-593.	0.7	12
54	Lack of apoptosis in patients with progressive external ophthalmoplegia and mutated adenine nucleotide translocator-1 gene. Muscle and Nerve, 2002, 26, 265-269.	1.0	12

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55	Schizophreniform Disorder with Cerebrospinal Fluid PCR Positivity for Herpes Simplex Virus Type 1. European Neurology, 2003, 50, 182-183.	0.6	12
56	Active intrathecal herpes simplex virus type 1 (HSV-1) and human herpesvirus-6 (HHV-6) infection at onset of multiple sclerosis. Journal of NeuroVirology, 2012, 18, 437-440.	1.0	12
57	Muscle manifestations and CK levels in COVID infection: results of a large cohort of patients inside a Pandemic COVID-19 Area. Acta Myologica, 2021, 40, 1-7.	1.5	12
58	Cytochrome c oxidase during human fetal development. International Journal of Developmental Neuroscience, 1989, 7, 5-14.	0.7	9
59	Duplication of dystrophin gene and dissimilar clinical phenotype in the same family. Neuromuscular Disorders, 1995, 5, 475-481.	0.3	8
60	Aphasic and visual aura with increased vasogenic leakage: An atypical migrainosus status. Journal of the Neurological Sciences, 2009, 285, 227-229.	0.3	8
61	Lack of anionic phospholipid calcium binding sites in duchenne muscular dystrophy. Muscle and Nerve, 1992, 15, 325-331.	1.0	7
62	Congenital muscular dystrophy with muscle inflammation alpha dystroglycan glycosylation defect and no mutation in FKRP gene. Journal of the Neurological Sciences, 2006, 243, 47-51.	0.3	6
63	Expression and localization of myotonic dystrophy protein kinase in human skeletal muscle cells determined with a novel antibody: Possible role of the protein in cytoskeleton rearrangements during differentiation. Cell Biology International, 2005, 29, 742-753.	1.4	5
64	Ryanodine receptor gene point mutation and malignant hyperthermia susceptibility. Journal of Neurology, 1995, 242, 127-133.	1.8	4
65	Severe polyneuropathy in a patient with Churg-Strauss syndrome. Journal of the Peripheral Nervous System, 2000, 5, 106-110.	1.4	3
66	Two dystrophin proteins and transcripts in a mild dystrophinopathic patient. Neuromuscular Disorders, 2003, 13, 13-16.	0.3	3
67	Is erythropoietin gene a modifier factor in amyotrophic lateral sclerosis?. Neurobiology of Aging, 2009, 30, 842-844.	1.5	3
68	Muscular Dystrophy: Central Nervous System Î±-Dystroglycan Glycosylation Defects and Brain Malformation. Journal of Child Neurology, 2010, 25, 312-320.	0.7	2
69	Anionic phospholipids calcium binding sites in Duchenne and murine X-linked muscular dystrophy. Muscle and Nerve, 1994, 17, 485-488.	1.0	1