Adele D amico

List of Publications by Citations

141 papers	5,218 citations	40 h-index	65 g-index
195 ext. papers	6,191 ext. citations	3 avg, IF	4.66 L-index

#	Paper	IF	Citations
141	Spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2011**, 6, 71	4.2	223
140	De novo LMNA mutations cause a new form of congenital muscular dystrophy. *Annals of Neurology*, **2008**, 64, 177-86	9.4	213
139	miRNAs as serum biomarkers for Duchenne muscular dystrophy. *EMBO Molecular Medicine*, **2011**, 3, 258-65	6.2	201
138	Gene expression profiling in the early phases of DMD: a constant molecular signature characterizes DMD muscle from early postnatal life throughout disease progression. *FASEB Journal*, **2007**, 21, 1210-26	10.9	168
137	Randomized, double-blind, placebo-controlled trial of phenylbutyrate in spinal muscular atrophy. *Neurology*, **2007**, 68, 51-5	6.5	141
136	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. *Brain*, **2007**, 130, 2024-36	11.2	138
135	North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2010**, 20, 712-6	2.9	134
134	Reliability of the North Star Ambulatory Assessment in a multicentric setting. *Neuromuscular Disorders*, **2009**, 19, 458-61	2.9	125
133	Leiomodin-3 dysfunction results in thin filament disorganization and nemaline myopathy. *Journal of Clinical Investigation*, **2014**, 124, 4693-708	15.9	118
132	Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. *Brain*, **2008**, 131, 747-59	11.2	112
131	Pilot trial of phenylbutyrate in spinal muscular atrophy. *Neuromuscular Disorders*, **2004**, 14, 130-5	2.9	109
130	Patterns of disease progression in type 2 and 3 SMA: Implications for clinical trials. *Neuromuscular Disorders*, **2016**, 26, 126-31	2.9	105
129	Histological effects of givinostat in boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2016**, 26, 643-649	2.9	96
128	GM1 gangliosidosis and Morquio B disease: an update on genetic alterations and clinical findings. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2011**, 1812, 782-90	6.9	95
127	Daily salbutamol in young patients with SMA type II. *Neuromuscular Disorders*, **2008**, 18, 536-40	2.9	83
126	Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36-month changes. *PLoS ONE*, **2014**, 9, e108205	3.7	82
125	Attention deficit hyperactivity disorder and cognitive function in Duchenne muscular dystrophy: phenotype-genotype correlation. *Journal of Pediatrics*, **2012**, 161, 705-9.e1	3.6	82

124	Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. *Neuromuscular Disorders*, **2010**, 20, 438-42	2.9	77
123	24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy. *PLoS ONE*, **2013**, 8, e52512	3.7	75
122	Natural history of pulmonary function in collagen VI-related myopathies. *Brain*, **2013**, 136, 3625-33	11.2	69
121	Reliability of the Performance of Upper Limb assessment in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2014**, 24, 201-6	2.9	68
120	Congenital muscular dystrophies: a brief review. *Seminars in Pediatric Neurology*, **2011**, 18, 277-88	2.9	68
119	Fatal hypertrophic cardiomyopathy and nemaline myopathy associated with ACTA1 K336E mutation. *Neuromuscular Disorders*, **2006**, 16, 548-52	2.9	68
118	Pontocerebellar hypoplasia type 6 caused by mutations in RARS2: definition of the clinical spectrum and molecular findings in five patients. *Journal of Inherited Metabolic Disease*, **2013**, 36, 43-53	5.4	62
117	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. *Neurology*, **2012**, 79, 159-62	6.5	62
116	Clinical and genetic characterization of Chanarin-Dorfman syndrome. *Biochemical and Biophysical Research Communications*, **2008**, 369, 1125-8	3.4	60
115	6 Minute walk test in Duchenne MD patients with different mutations: 12 month changes. *PLoS ONE*, **2014**, 9, e83400	3.7	56
114	Salbutamol increases survival motor neuron (SMN) transcript levels in leucocytes of spinal muscular atrophy (SMA) patients: relevance for clinical trial design. *Journal of Medical Genetics*, **2010**, 47, 856-8	5.8	54
113	Two patients with SDropped head syndromeSdue to mutations in LMNA or SEPN1 genes. *Neuromuscular Disorders*, **2005**, 15, 521-4	2.9	51
112	Reliability of the Hammersmith functional motor scale for spinal muscular atrophy in a multicentric study. *Neuromuscular Disorders*, **2006**, 16, 93-8	2.9	51
111	Expanding the clinical spectrum of POMT1 phenotype. *Neurology*, **2006**, 66, 1564-7; discussion 1461	6.5	50
110	Prevalence of congenital muscular dystrophy in Italy: a population study. *Neurology*, **2015**, 84, 904-11	6.5	49
109	Functional and Morphological Improvement of Dystrophic Muscle by Interleukin 6 Receptor Blockade. *EBioMedicine*, **2015**, 2, 285-93	8.8	47
108	Long term follow-up to evaluate the efficacy of miglustat treatment in Italian patients with Niemann-Pick disease type C. *Orphanet Journal of Rare Diseases*, **2015**, 10, 22	4.2	45
107	Centronuclear myopathy related to dynamin 2 mutations: clinical, morphological, muscle imaging and genetic features of an Italian cohort. *Neuromuscular Disorders*, **2013**, 23, 229-38	2.9	45

106	LMNA-associated myopathies: the Italian experience in a large cohort of patients. *Neurology*, **2014**, 83, 1634-44	6.5	45
105	Centronuclear myopathies: genotype-phenotype correlation and frequency of defined genetic forms in an Italian cohort. *Journal of Neurology*, **2015**, 262, 1728-40	5.5	42
104	Duchenne muscular dystrophy and epilepsy. *Neuromuscular Disorders*, **2013**, 23, 313-5	2.9	42
103	Genetic characterization in symptomatic female DMD carriers: lack of relationship between X-inactivation, transcriptional DMD allele balancing and phenotype. *BMC Medical Genetics*, **2012**, 13, 73	2.1	42
102	Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations. *Journal of Neurology*, **2014**, 261, 870-6	5.5	41
101	The Hammersmith functional score correlates with the SMN2 copy number: a multicentric study. *Neuromuscular Disorders*, **2007**, 17, 400-3	2.9	39
100	Exon 45 skipping through U1-snRNA antisense molecules recovers the Dys-nNOS pathway and muscle differentiation in human DMD myoblasts. *Molecular Therapy*, **2012**, 20, 2134-42	11.7	37
99	TBCE Mutations Cause Early-Onset Progressive Encephalopathy with Distal Spinal Muscular Atrophy. *American Journal of Human Genetics*, **2016**, 99, 974-983	11	37
98	A novel AIFM1 mutation expands the phenotype to an infantile motor neuron disease. *European Journal of Human Genetics*, **2016**, 24, 463-6	5.3	36
97	SMN transcript levels in leukocytes of SMA patients determined by absolute real-time PCR. *European Journal of Human Genetics*, **2010**, 18, 52-8	5.3	36
96	Benefits of glucocorticoids in non-ambulant boys/men with Duchenne muscular dystrophy: A multicentric longitudinal study using the Performance of Upper Limb test. *Neuromuscular Disorders*, **2015**, 25, 749-53	2.9	35
95	Six minute walk test in type III spinal muscular atrophy: a 12month longitudinal study. *Neuromuscular Disorders*, **2013**, 23, 624-8	2.9	35
94	POMT2 mutation in a patient with SMEB-likeSphenotype. *Neuromuscular Disorders*, **2006**, 16, 446-8	2.9	35
93	Neutral Lipid Storage Diseases: clinical/genetic features and natural history in a large cohort of Italian patients. *Orphanet Journal of Rare Diseases*, **2017**, 12, 90	4.2	34
92	Increased muscle expression of interleukin-17 in Duchenne muscular dystrophy. *Neurology*, **2012**, 78, 1309-14	6.5	34
91	X-linked myotubular myopathy: A prospective international natural history study. *Neurology*, **2019**, 92, e1852-e1867	6.5	33
90	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPT-II deficiency. *Clinical Genetics*, **2012**, 82, 232-9	4	33
89	Molecular analysis of NPC1 and NPC2 gene in 34 Niemann-Pick C Italian patients: identification and structural modeling of novel mutations. *Neurogenetics*, **2009**, 10, 229-39	3	33

88	Hammersmith Functional Motor Scale and Motor Function Measure-20 in non ambulant SMA patients. *Neuromuscular Disorders*, **2014**, 24, 347-52	2.9	32
87	Brown-Vialetto-van Laere and Fazio-Londe overlap syndromes: a clinical, biochemical and genetic study. *Neuromuscular Disorders*, **2012**, 22, 1075-82	2.9	32
86	POMGnT1 mutations in congenital muscular dystrophy: genotype-phenotype correlation and expanded clinical spectrum. *Archives of Neurology*, **2006**, 63, 1491-5		30
85	Histologic muscular history in steroid-treated and untreated patients with Duchenne dystrophy. *Neurology*, **2015**, 85, 1886-93	6.5	29
84	Identification and characterization of novel collagen VI non-canonical splicing mutations causing Ullrich congenital muscular dystrophy. *Human Mutation*, **2009**, 30, E662-72	4.7	29
83	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. *PLoS ONE*, **2018**, 13, e0199223	3.7	28
82	Persistent pulmonary arterial hypertension in the newborn (PPHN): a frequent manifestation of TMEM70 defective patients. *Molecular Genetics and Metabolism*, **2014**, 111, 353-359	3.7	28
81	POMT1 and POMT2 mutations in CMD patients: a multicentric Italian study. *Neuromuscular Disorders*, **2008**, 18, 565-71	2.9	28
80	Genomic rearrangements at the IGHMBP2 gene locus in two patients with SMARD1. *Human Genetics*, **2004**, 115, 319-26	6.3	28
79	Riboflavin transporter 3 involvement in infantile Brown-Vialetto-Van Laere disease: two novel mutations. *Journal of Medical Genetics*, **2013**, 50, 104-7	5.8	27
78	Revised North Star Ambulatory Assessment for Young Boys with Duchenne Muscular Dystrophy. *PLoS ONE*, **2016**, 11, e0160195	3.7	27
77	The use of muscle biopsy in the diagnosis of undefined ataxia with cerebellar atrophy in children. *European Journal of Paediatric Neurology*, **2012**, 16, 248-56	3.8	26
76	Respiratory and cardiac function in congenital muscular dystrophies with alpha dystroglycan deficiency. *Neuromuscular Disorders*, **2012**, 22, 685-9	2.9	26
75	Spinal muscular atrophy associated with progressive myoclonic epilepsy: A rare condition caused by mutations in ASAH1. *Epilepsia*, **2015**, 56, 692-8	6.4	25
74	Heart transplantation in a child with LGMD2I presenting as isolated dilated cardiomyopathy. *Neuromuscular Disorders*, **2008**, 18, 153-5	2.9	25
73	Diagnosis of Duchenne Muscular Dystrophy in Italy in the last decade: Critical issues and areas for improvements. *Neuromuscular Disorders*, **2017**, 27, 447-451	2.9	24
72	Suitability of North Star Ambulatory Assessment in young boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2015**, 25, 14-8	2.9	24
71	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. *European Journal of Human Genetics*, **2012**, 20, 1234-9	5.3	23

70	Clinical-genetic features and peculiar muscle histopathology in infantile DNM1L-related mitochondrial epileptic encephalopathy. *Human Mutation*, **2019**, 40, 601-618	4.7	22
69	Congenital myopathies. *Current Neurology and Neuroscience Reports*, **2008**, 8, 73-9	6.6	22
68	Hypertrophic cardiomyopathy, cataract, developmental delay, lactic acidosis: a novel subtype of 3-methylglutaconic aciduria. *Journal of Inherited Metabolic Disease*, **2006**, 29, 546-50	5.4	22
67	Phenotypic heterogeneity in two unrelated Danon patients associated with the same LAMP-2 gene mutation. *Neuropediatrics*, **2005**, 36, 309-13	1.6	22
66	Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study. *PLoS ONE*, **2016**, 11, e0151445	3.7	22
65	Cardiac and Neuromuscular Features of Patients With LMNA-Related Cardiomyopathy. *Annals of Internal Medicine*, **2019**, 171, 458-463	8	22
64	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. *PLoS ONE*, **2019**, 14, e0218683	3.7	21
63	The 6 minute walk test and performance of upper limb in ambulant duchenne muscular dystrophy boys. *PLOS Currents*, **2014**, 6,		21
62	Broad phenotypic spectrum and genotype-phenotype correlations in GMPPB-related dystroglycanopathies: an Italian cross-sectional study. *Orphanet Journal of Rare Diseases*, **2018**, 13, 170	4.2	21
61	Novel findings associated with MTM1 suggest a higher number of female symptomatic carriers. *Neuromuscular Disorders*, **2016**, 26, 292-9	2.9	20
60	Health-related quality of life and functional changes in DMD: A 12-month longitudinal cohort study. *Neuromuscular Disorders*, **2016**, 26, 189-96	2.9	20
59	Ullrich myopathy phenotype with secondary ColVI defect identified by confocal imaging and electron microscopy analysis. *Neuromuscular Disorders*, **2007**, 17, 587-96	2.9	20
58	Effect of mexiletine on transitory depression of compound motor action potential in recessive myotonia congenita. *Clinical Neurophysiology*, **2015**, 126, 399-403	4.3	18
57	Development of an academic disease registry for spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, 794-799	2.9	17
56	Low-rate repetitive nerve stimulation protocol in an Italian cohort of patients affected by recessive myotonia congenita. *Journal of Clinical Neurophysiology*, **2011**, 28, 39-44	2.2	17
55	Insulin-like growth factor I in inclusion-body myositis and human muscle cultures. *Journal of Neuropathology and Experimental Neurology*, **2004**, 63, 650-9	3.1	17
54	Efficacy of miglustat in Niemann-Pick C disease: a single centre experience. *Molecular Genetics and Metabolism*, **2013**, 110, 329-35	3.7	16
53	Early Neurodevelopmental Findings Predict School Age Cognitive Abilities in Duchenne Muscular Dystrophy: A Longitudinal Study. *PLoS ONE*, **2015**, 10, e0133214	3.7	16

52	Major myofibrillar changes in early onset myopathy due to de novo heterozygous missense mutation in lamin A/C gene. *Neuromuscular Disorders*, **2005**, 15, 847-50	2.9	16
51	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. *Neurological Sciences*, **2019**, 40, 457-468	3.5	16
50	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. *PLoS ONE*, **2020**, 15, e0230677	3.7	15
49	Rituximab in a childhood-onset idiopathic refractory chronic inflammatory demyelinating polyneuropathy. *European Journal of Paediatric Neurology*, **2012**, 16, 301-3	3.8	15
48	A new de novo missense mutation in MYH2 expands clinical and genetic findings in hereditary myosin myopathies. *Neuromuscular Disorders*, **2013**, 23, 437-40	2.9	15
47	Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. *Neuromuscular Disorders*, **2011**, 21, 194-203	2.9	15
46	Translational approach to address therapy in myotonia permanens due to a new SCN4A mutation. *Neurology*, **2016**, 86, 2100-8	6.5	15
45	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 786-798	5.3	14
44	Clinical Variability in Spinal Muscular Atrophy Type III. *Annals of Neurology*, **2020**, 88, 1109-1117	9.4	14
43	Implicit learning deficit in children with Duchenne muscular dystrophy: Evidence for a cerebellar cognitive impairment?. *PLoS ONE*, **2018**, 13, e0191164	3.7	13
42	Fetal acetylcholine receptor inactivation syndrome and maternal myasthenia gravis: a case report. *Neuromuscular Disorders*, **2012**, 22, 546-8	2.9	13
41	New Mutations in NEB Gene Discovered by Targeted Next-Generation Sequencing in Nemaline Myopathy Italian Patients. *Journal of Molecular Neuroscience*, **2016**, 59, 351-9	3.3	13
40	An observational study of functional abilities in infants, children, and adults with type 1 SMA. *Neurology*, **2018**, 91, e696-e703	6.5	12
39	Longitudinal natural history in young boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2019**, 29, 857-862	2.9	12
38	Deep RNA profiling identified CLOCK and molecular clock genes as pathophysiological signatures in collagen VI myopathy. *Journal of Cell Science*, **2016**, 129, 1671-84	5.3	12
37	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. *Neurology*, **2021**, 96, e587-e599	6.5	12
36	Muscle MRI in neutral lipid storage disease (NLSD). *Journal of Neurology*, **2017**, 264, 1334-1342	5.5	11
35	Muscle magnetic resonance imaging and histopathology in ACTA1-related congenital nemaline myopathy. *Muscle and Nerve*, **2014**, 50, 1011-6	3.4	11

34	Duchenne Dilated Cardiomyopathy: Cardiac Management from Prevention to Advanced Cardiovascular Therapies. *Journal of Clinical Medicine*, **2020**, 9,	5.1	10
33	Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. *International Journal of Cardiology*, **2019**, 280, 99-103	3.2	9
32	Cardiac function in types II and III spinal muscular atrophy: should we change standards of care?. *Neuropediatrics*, **2015**, 46, 33-6	1.6	9
31	Characterization of a rare case of Ullrich congenital muscular dystrophy due to truncating mutations within the COL6A1 gene C-terminal domain: a case report. *BMC Medical Genetics*, **2013**, 14, 59	2.1	9
30	Muscle imaging in patients with tubular aggregate myopathy caused by mutations in STIM1. *Neuromuscular Disorders*, **2015**, 25, 898-903	2.9	9
29	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. *Neuromuscular Disorders*, **2021**, 31, 596-602	2.9	9
28	Long-Term Safety and Usefulness of Mexiletine in a Large Cohort of Patients Affected by Non-dystrophic Myotonias. *Frontiers in Neurology*, **2020**, 11, 300	4.1	8
27	SAmish Nemaline MyopathySin 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. *Neuromuscular Disorders*, **2019**, 29, 766-770	2.9	8
26	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1622-1634	5.3	6
25	Developmental lag of visuospatial attention in Duchenne muscular dystrophy. *Research in Developmental Disabilities*, **2015**, 36C, 55-61	2.7	5
24	Old measures and new scores in spinal muscular atrophy patients. *Muscle and Nerve*, **2015**, 52, 435-7	3.4	5
23	The empowerment of translational research: lessons from laminopathies. *Orphanet Journal of Rare Diseases*, **2012**, 7, 37	4.2	5
22	Spinal muscular atrophy: state of the art and new therapeutic strategies. *Neurological Sciences*, **2021**, 1	3.5	5
21	Predictive energy equations for spinal muscular atrophy type I children. *American Journal of Clinical Nutrition*, **2020**, 111, 983-996	7	4
20	Evaluation of gait in Duchenne Muscular Dystrophy: Relation of 3D gait analysis to clinical assessment. *Neuromuscular Disorders*, **2019**, 29, 920-929	2.9	4
19	Somatic mosaicism represents an underestimated event underlying collagen 6-related disorders. *European Journal of Paediatric Neurology*, **2017**, 21, 873-883	3.8	4
18	A new form of alpha-dystroglycanopathy associated with severe drug-resistant epilepsy and unusual EEG features. *Epileptic Disorders*, **2011**, 13, 259-62	1.9	4
17	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. *Genes*, **2018** , 9,	4.2	4

16	Mitochondrial encephalomyopathies and related syndromes: brief review. *Cerebrovascular Diseases*, **2009**, 14, 38-52	3.2	3
15	Neonatal-onset nemaline myopathy mimicking congenital diaphragmatic hernia. *Journal of Pediatric Surgery*, **2007**, 42, E19-22	2.6	3
14	Growth patterns in children with spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2021**, 16, 375	4.2	3
13	Cytokine Profile in Striated Muscle Laminopathies: New Promising Biomarkers for Disease Prediction. *Cells*, **2020**, 9,	7.9	2
12	Integrated care of muscular dystrophies in Italy. Part 1. Pharmacological treatment and rehabilitative interventions. *Acta Myologica*, **2017**, 36, 19-24	1.6	2
11	Hepatobiliary disease in XLMTM: a common comorbidity with potential impact on treatment strategies. *Orphanet Journal of Rare Diseases*, **2021**, 16, 425	4.2	2
10	A Recurrent Pathogenic Variant of Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. *Frontiers in Genetics*, **2020**, 11, 565868	4.5	2
9	International retrospective natural history study of -related congenital muscular dystrophy. *Brain Communications*, **2021**, 3, fcab075	4.5	2
8	Muscle imaging in fibrodysplasia ossificans progressiva: The neurologistS perspective. *Neuromuscular Disorders*, **2015**, 25, 672-3	2.9	1
7	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. *PLoS ONE*, **2021**, 16, e0253882	3.7	1
6	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study.. *Acta Neuropathologica Communications*, **2022**, 10, 54	7.3	1
5	Genetic modifiers of upper limb function in Duchenne muscular dystrophy.. *Journal of Neurology*, **2022**, 1	5.5	1
4	Body mass index in type 2 spinal muscular atrophy: a longitudinal study.. *European Journal of Pediatrics*, **2022**, 1	4.1	0
3	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the Mouse Model and Patients. *Frontiers in Physiology*, **2021**, 12, 678974	4.6	0
2	Acute quadriplegic myopathy in a 16-month-old child. *Paediatric Anaesthesia*, **2005**, 15, 611-5	1.8	
1	Age-related sensory neuropathy in patients with spinal muscular atrophy type 1. *Muscle and Nerve*, **2021**, 64, 599-603	3.4	