Corrado Angelini

List of Publications by Citations

569 papers	21,484 citations	75 h-index	119 g-index
655 ext. papers	23,836 ext. citations	5.2 avg, IF	6.4 L-index

#	Paper	IF	Citations
569	Dysferlin, a novel skeletal muscle gene, is mutated in Miyoshi myopathy and limb girdle muscular dystrophy. *Nature Genetics*, **1998**, 20, 31-6	36.3	735
568	Assessment of mitochondrial respiratory chain enzymatic activities on tissues and cultured cells. *Nature Protocols*, **2012**, 7, 1235-46	18.8	539
567	MELAS: clinical features, biochemistry, and molecular genetics. *Annals of Neurology*, **1992**, 31, 391-8	9.4	456
566	MELAS mutation in mtDNA binding site for transcription termination factor causes defects in protein synthesis and in respiration but no change in levels of upstream and downstream mature transcripts. *Proceedings of the National Academy of Sciences of the United States of America*, **1992**, 89, 4221-5	11.5	455
565	Carnitine deficiency of human skeletal muscle with associated lipid storage myopathy: a new syndrome. *Science*, **1973**, 179, 899-902	33.3	437
564	Beta-sarcoglycan (A3b) mutations cause autosomal recessive muscular dystrophy with loss of the sarcoglycan complex. *Nature Genetics*, **1995**, 11, 266-73	36.3	405
563	Gene for chronic proximal spinal muscular atrophies maps to chromosome 5q. *Nature*, **1990**, 344, 767-8	50.4	343
562	Exploring the molecular basis for variability among patients with Becker muscular dystrophy: dystrophin gene and protein studies. *American Journal of Human Genetics*, **1991**, 49, 54-67	11	281
561	Safety and efficacy of eculizumab in anti-acetylcholine receptor antibody-positive refractory generalised myasthenia gravis (REGAIN): a phase 3, randomised, double-blind, placebo-controlled, multicentre study. *Lancet Neurology, The*, **2017**, 16, 976-986	24.1	278
560	Nuclear envelope dystrophies show a transcriptional fingerprint suggesting disruption of Rb-MyoD pathways in muscle regeneration. *Brain*, **2006**, 129, 996-1013	11.2	250
559	Improved diagnosis of Becker muscular dystrophy by dystrophin testing. *Neurology*, **1989**, 39, 1011-7	6.5	217
558	Systemic carnitine deficiency--a treatable inherited lipid-storage disease presenting as Reye's syndrome. *New England Journal of Medicine*, **1980**, 303, 1389-94	59.2	215
557	Mutations in the sarcoglycan genes in patients with myopathy. *New England Journal of Medicine*, **1997**, 336, 618-24	59.2	196
556	Correlating phenotype and genotype in the periodic paralyses. *Neurology*, **2004**, 63, 1647-55	6.5	177
555	Cerebellar ataxia and coenzyme Q10 deficiency. *Neurology*, **2003**, 60, 1206-8	6.5	174
554	A multicenter, double-blind, randomized trial of deflazacort versus prednisone in Duchenne muscular dystrophy. *Muscle and Nerve*, **2000**, 23, 1344-7	3.4	170
553	Myotonic dystrophy: evidence for a possible dominant-negative RNA mutation. *Human Molecular Genetics*, **1995**, 4, 599-606	5.6	169

552	Survival motor neuron gene transcript analysis in muscles from spinal muscular atrophy patients. *Biochemical and Biophysical Research Communications*, **1995**, 213, 342-8	3.4	169
551	Infantile encephalomyopathy and nephropathy with CoQ10 deficiency: a CoQ10-responsive condition. *Neurology*, **2005**, 65, 606-8	6.5	168
550	SPP1 genotype is a determinant of disease severity in Duchenne muscular dystrophy. *Neurology*, **2011**, 76, 219-26	6.5	150
549	A point mutation in the 5' splice site of the dystrophin gene first intron responsible for X-linked dilated cardiomyopathy. *Human Molecular Genetics*, **1996**, 5, 73-9	5.6	147
548	Increases in walking distance in patients with peripheral vascular disease treated with L-carnitine: a double-blind, cross-over study. *Circulation*, **1988**, 77, 767-73	16.7	143
547	Carnitine deficiency, organic acidemias, and Reye's syndrome. *Neurology*, **1985**, 35, 1041-5	6.5	139
546	A stop-codon mutation in the human mtDNA cytochrome c oxidase I gene disrupts the functional structure of complex IV. *American Journal of Human Genetics*, **1999**, 65, 611-20	11	138
545	Disruption of skeletal muscle mitochondrial network genes and miRNAs in amyotrophic lateral sclerosis. *Neurobiology of Disease*, **2013**, 49, 107-17	7.5	137
544	The role of corticosteroids in muscular dystrophy: a critical appraisal. *Muscle and Nerve*, **2007**, 36, 424-35	3.4	137
543	Myocardial involvement is very frequent among patients affected with subclinical Becker's muscular dystrophy. *Circulation*, **1996**, 94, 3168-75	16.7	137
542	Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation. *Neurology*, **2013**, 80, 2049-54	6.5	135
541	Deflazacort in Duchenne dystrophy: study of long-term effect. *Muscle and Nerve*, **1994**, 17, 386-91	3.4	130
540	Reliability of the North Star Ambulatory Assessment in a multicentric setting. *Neuromuscular Disorders*, **2009**, 19, 458-61	2.9	125
539	Dystrophinopathy in isolated cases of myopathy in females. *Neurology*, **1992**, 42, 967-75	6.5	114
538	Spinal and bulbar muscular atrophy: skeletal muscle pathology in male patients and heterozygous females. *Journal of the Neurological Sciences*, **2008**, 264, 100-5	3.2	109
537	Human skeletal muscle atrophy in amyotrophic lateral sclerosis reveals a reduction in Akt and an increase in atrogin-1. *FASEB Journal*, **2006**, 20, 583-5	0.9	109
536	Muscle MRI findings in patients with limb girdle muscular dystrophy with calpain 3 deficiency (LGMD2A) and early contractures. *Neuromuscular Disorders*, **2005**, 15, 164-71	2.9	108
535	Genomic organization of the dysferlin gene and novel mutations in Miyoshi myopathy. *Neurology*, **2001**, 57, 271-8	6.5	108

534	MtDNA mutations associated with Leber's hereditary optic neuropathy: studies on cytoplasmic hybrid (cybrid) cells. *Biochemical and Biophysical Research Communications*, **1995**, 210, 880-8	3.4	108
533	Laminin alpha2 muscular dystrophy: genotype/phenotype studies of 22 patients. *Neurology*, **1998**, 51, 101-10	6.5	107
532	Cardiac involvement in Becker muscular dystrophy. *Journal of the American College of Cardiology*, **1993**, 22, 1927-34	15.1	106
531	Cardiac and respiratory involvement in advanced stage Duchenne muscular dystrophy. *Neuromuscular Disorders*, **1996**, 6, 367-76	2.9	105
530	EFNS guidelines on the diagnostic approach to pauci- or asymptomatic hyperCKemia. *European Journal of Neurology*, **2010**, 17, 767-73	6	104
529	Ubidecarenone in the treatment of mitochondrial myopathies: a multi-center double-blind trial. *Journal of the Neurological Sciences*, **1990**, 100, 70-8	3.2	103
528	Mutations that disrupt the carboxyl-terminus of gamma-sarcoglycan cause muscular dystrophy. *Human Molecular Genetics*, **1996**, 5, 1841-7	5.6	102
527	Caspase 3 expression correlates with skeletal muscle apoptosis in Duchenne and facioscapulo human muscular dystrophy. A potential target for pharmacological treatment?. *Journal of Neuropathology and Experimental Neurology*, **2001**, 60, 302-12	3.1	99
526	Correlation between cardiac involvement and CTG trinucleotide repeat length in myotonic dystrophy. *Journal of the American College of Cardiology*, **1995**, 25, 239-45	15.1	98
525	Genetic epidemiology of congenital muscular dystrophy in a sample from north-east Italy. *Human Genetics*, **1996**, 97, 277-9	6.3	97
524	Regulation of ER-mitochondria contacts by Parkin via Mfn2. *Pharmacological Research*, **2018**, 138, 43-56	10.2	97
523	Muscle pathology in dysferlin deficiency. *Neuropathology and Applied Neurobiology*, **2002**, 28, 461-70	5.2	96
522	Identification of 45 novel mutations in the nebulin gene associated with autosomal recessive nemaline myopathy. *Human Mutation*, **2006**, 27, 946-56	4.7	95
521	Heart involvement in muscular dystrophies due to sarcoglycan gene mutations. *Muscle and Nerve*, **1999**, 22, 473-9	3.4	95
520	Observational clinical study in juvenile-adult glycogenosis type 2 patients undergoing enzyme replacement therapy for up to 4 years. *Journal of Neurology*, **2012**, 259, 952-8	5.5	93
519	A multicentre follow-up study of 1152 patients with myasthenia gravis in Italy. *Journal of Neurology* , **1990**, 237, 339-44	5.5	93
518	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. *Journal of Neurology*, **2014**, 261, 504-10	5.5	91
517	Population frequency of myotonic dystrophy: higher than expected frequency of myotonic dystrophy type 2 (DM2) mutation in Finland. *European Journal of Human Genetics*, **2011**, 19, 776-82	5.3	90

516	Molecular diagnosis in LGMD2A: mutation analysis or protein testing?. *Human Mutation*, **2004**, 24, 52-62	4.7	90
515	Facioscapulohumeral muscular dystrophy: epidemiological and molecular study in a north-east Italian population sample. *Clinical Genetics*, **2009**, 75, 550-5	4	88
514	Carnitine deficiency of skeletal muscle: report of a treated case. *Neurology*, **1976**, 26, 633-7	6.5	88
513	Large-scale population analysis challenges the current criteria for the molecular diagnosis of fascioscapulohumeral muscular dystrophy. *American Journal of Human Genetics*, **2012**, 90, 628-35	11	86
512	Overexpression of microRNA-206 in the skeletal muscle from myotonic dystrophy type 1 patients. *Journal of Translational Medicine*, **2010**, 8, 48	8.5	86
511	Phenotype modulators in myophosphorylase deficiency. *Annals of Neurology*, **2003**, 53, 497-502	9.4	86
510	Is the carboxyl-terminus of dystrophin required for membrane association? A novel, severe case of Duchenne muscular dystrophy. *Annals of Neurology*, **1991**, 30, 605-10	9.4	86
509	A standardized clinical evaluation of patients affected by facioscapulohumeral muscular dystrophy: The FSHD clinical score. *Muscle and Nerve*, **2010**, 42, 213-7	3.4	84
508	Comparative study of acid maltase deficiency. Biochemical differences between infantile, childhood, and adult types. *Archives of Neurology*, **1972**, 26, 344-9		84
507	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience. *European Journal of Neurology*, **2017**, 24, 768-e31	6	81
506	TARDBP (TDP-43) sequence analysis in patients with familial and sporadic ALS: identification of two novel mutations. *European Journal of Neurology*, **2009**, 16, 727-32	6	81
505	A novel deletion in the GTPase domain of OPA1 causes defects in mitochondrial morphology and distribution, but not in function. *Human Molecular Genetics*, **2008**, 17, 3291-302	5.6	80
504	Tubular aggregates: sarcoplasmic reticulum origin, calcium storage ability, and functional implications. *Muscle and Nerve*, **1985**, 8, 299-306	3.4	80
503	Role of gabapentin in spinal muscular atrophy: results of a multicenter, randomized Italian study. *Journal of Child Neurology*, **2003**, 18, 537-41	2.5	79
502	Evaluation of muscle biopsy in late-onset GSDII patients before and after enzyme replacement therapy (ERT). *BMC Musculoskeletal Disorders*, **2013**, 14, P13	2.8	78
501	Brain involvement in myotonic dystrophies: neuroimaging and neuropsychological comparative study in DM1 and DM2. *Journal of Neurology*, **2010**, 257, 1246-55	5.5	78
500	Genotype-phenotype correlations of DHP receptor alpha 1-subunit gene mutations causing hypokalemic periodic paralysis. *Neuromuscular Disorders*, **1997**, 7, 33-8	2.9	78
499	The role of autophagy in the pathogenesis of glycogen storage disease type II (GSDII). *Cell Death and Differentiation*, **2012**, 19, 1698-708	12.7	77

498	Genetic and biochemical normalization in female carriers of Duchenne muscular dystrophy: evidence for failure of dystrophin production in dystrophin-competent myonuclei. *Neurology*, **1995**, 45, 677-90	6.5	77
497	Loss of calpain-3 autocatalytic activity in LGMD2A patients with normal protein expression. *American Journal of Pathology*, **2003**, 163, 1929-36	5.8	76
496	Ultrastructural changes in dysferlinopathy support defective membrane repair mechanism. *Journal of Clinical Pathology*, **2005**, 58, 190-5	3.9	76
495	Decorin and biglycan expression is differentially altered in several muscular dystrophies. *Brain*, **2005**, 128, 2546-55	11.2	75
494	Extensive scanning of the calpain-3 gene broadens the spectrum of LGMD2A phenotypes. *Journal of Medical Genetics*, **2005**, 42, 686-93	5.8	74
493	Coenzyme Q10 is frequently reduced in muscle of patients with mitochondrial myopathy. *Neuromuscular Disorders*, **2010**, 20, 44-8	2.9	73
492	The clinical spectrum of sarcoglycanopathies. *Neurology*, **1999**, 52, 176-9	6.5	72
491	Large scale genotype-phenotype analyses indicate that novel prognostic tools are required for families with facioscapulohumeral muscular dystrophy. *Brain*, **2013**, 136, 3408-17	11.2	71
490	Frequency of LGMD gene mutations in Italian patients with distinct clinical phenotypes. *Neurology*, **2009**, 72, 1432-5	6.5	71
489	The genetic basis of undiagnosed muscular dystrophies and myopathies: Results from 504 patients. *Neurology*, **2016**, 87, 71-6	6.5	70
488	The frequency of limb girdle muscular dystrophy 2A in northeastern Italy. *Neuromuscular Disorders*, **2005**, 15, 218-24	2.9	70
487	Riboflavin therapy. Biochemical heterogeneity in two adult lipid storage myopathies. *Brain*, **1999**, 122 (Pt 12), 2401-11	11.2	70
486	Multisystem triglyceride storage disorder with impaired long-chain fatty acid oxidation. *Annals of Neurology*, **1980**, 7, 5-10	9.4	70
485	Carnitine, carnitine acyltransferases, and rat brain function. *Experimental Neurology*, **1982**, 78, 285-92	5.7	70
484	Clinical and molecular characterization of patients with limb-girdle muscular dystrophy type 2I. *Archives of Neurology*, **2005**, 62, 1894-9		68
483	Exploring mental status in Friedreich's ataxia: a combined neuropsychological, behavioral and neuroimaging study. *European Journal of Neurology*, **2006**, 13, 827-35	6	66
482	The role of botulinum toxin injection and upper esophageal sphincter myotomy in treating oropharyngeal dysphagia. *Journal of Gastrointestinal Surgery*, **2004**, 8, 997-1006	3.3	65
481	Generalized lysosome-associated membrane protein-2 defect explains multisystem clinical involvement and allows leukocyte diagnostic screening in Danon disease. *American Journal of Pathology*, **2006**, 168, 1309-20	5.8	64

480	Investigating the Mechanism of Chromosomal Deletion: Characterization of 39 Deletion Breakpoints in Introns 47 and 48 of the Human Dystrophin Gene. *Genomics*, **2002**, 80, 523-530	4.3	64
479	Genetic epidemiology of muscular dystrophies resulting from sarcoglycan gene mutations. *Journal of Medical Genetics*, **1997**, 34, 973-7	5.8	63
478	Homozygous alpha-sarcoglycan mutation in two siblings: one asymptomatic and one steroid-responsive mild limb-girdle muscular dystrophy patient. *Muscle and Nerve*, **1998**, 21, 769-75	3.4	63
477	Motor function-muscle strength relationship in spinal muscular atrophy. *Muscle and Nerve*, **2004**, 29, 548-52	3.4	63
476	Gene expression profiling in dysferlinopathies using a dedicated muscle microarray. *Human Molecular Genetics*, **2002**, 11, 3283-98	5.6	63
475	Muscle carnitine deficiency in patients with severe peripheral vascular disease. *Circulation*, **1991**, 84, 1490-5	16.7	63
474	Importance of SPP1 genotype as a covariate in clinical trials in Duchenne muscular dystrophy. *Neurology*, **2012**, 79, 159-62	6.5	62
473	Normal myogenesis and increased apoptosis in myotonic dystrophy type-1 muscle cells. *Cell Death and Differentiation*, **2010**, 17, 1315-24	12.7	61
472	Memory deficits and retrieval processes in ALS. *European Journal of Neurology*, **2003**, 10, 221-7	6	61
471	Carnitine deficiency induced during hemodialysis and hyperlipidemia: effect of replacement therapy. *American Journal of Clinical Nutrition*, **1981**, 34, 1496-500	7	61
470	Clinical and genetic characterization of Chanarin-Dorfman syndrome. *Biochemical and Biophysical Research Communications*, **2008**, 369, 1125-8	3.4	60
469	Type 1 fiber abnormalities in skeletal muscle of patients with hypertrophic and dilated cardiomyopathy: evidence of subclinical myogenic myopathy. *Journal of the American College of Cardiology*, **1989**, 14, 1464-73	15.1	59
468	MRI in the assessment of muscular pathology: a comparison between limb-girdle muscular dystrophies, hyaline body myopathies and myotonic dystrophies. *Radiologia Medica*, **2010**, 115, 585-99	6.5	58
467	Calpain-3 and dysferlin protein screening in patients with limb-girdle dystrophy and myopathy. *Neurology*, **2001**, 56, 660-5	6.5	58
466	Dystrophin-positive fibers in Duchenne dystrophy: origin and correlation to clinical course. *Muscle and Nerve*, **1995**, 18, 1115-20	3.4	58
465	Molecular and muscle pathology in a series of caveolinopathy patients. *Human Mutation*, **2005**, 25, 82-9	4.7	57
464	Integrin alpha 7 beta 1 in muscular dystrophy/myopathy of unknown etiology. *American Journal of Pathology*, **2002**, 160, 2135-43	5.8	57
463	Cognitive impairment and (CTG)n expansion in myotonic dystrophy patients. *Biological Psychiatry*, **1999**, 46, 425-31	7.9	57

462	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. *Muscle and Nerve*, **2017**, 55, 55-68	3.4	56
461	Myotonia and the muscle chloride channel: dominant mutations show variable penetrance and founder effect. *Neurology*, **1996**, 47, 963-8	6.5	56
460	Sensory, motor, and autonomic neuropathy in patients with multiple symmetric lipomatosis. *Medicine (United States)*, **1985**, 64, 388-93	1.8	56
459	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. *Neuromuscular Disorders*, **2015**, 25, 533-41	2.9	55
458	Dominant LMNA mutations can cause combined muscular dystrophy and peripheral neuropathy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2005**, 76, 1019-21	5.5	55
457	Redefining phenotypes associated with mitochondrial DNA single deletion. *Journal of Neurology*, **2015**, 262, 1301-9	5.5	53
456	Next-generation sequencing identifies transportin 3 as the causative gene for LGMD1F. *PLoS ONE*, **2013**, 8, e63536	3.7	53
455	Muscular dystrophy with marked Dysferlin deficiency is consistently caused by primary dysferlin gene mutations. *European Journal of Human Genetics*, **2011**, 19, 974-80	5.3	53
454	Facioscapulohumeral muscular dystrophy and occurrence of heart arrhythmia. *European Neurology*, **2006**, 56, 1-5	2.1	53
453	Mitochondria-lipid-glycogen (MLG) disease of muscle. A morphologically regressive congenital myopathy. *Archives of Neurology*, **1973**, 29, 162-9		53
452	RNA metabolism in myotonic dystrophy: patient muscle shows decreased insulin receptor RNA and protein consistent with abnormal insulin resistance. *Journal of Clinical Investigation*, **1997**, 99, 1691-8	15.9	53
451	Circulating microRNAs as biomarkers of muscle differentiation and atrophy in ALS **2016**, 35, 22-30		53
450	Old and new therapeutic developments in steroid treatment in Duchenne muscular dystrophy. *Acta Myologica*, **2012**, 31, 9-15	1.6	52
449	Different atrophy-hypertrophy transcription pathways in muscles affected by severe and mild spinal muscular atrophy. *BMC Medicine*, **2009**, 7, 14	11.4	51
448	Long-term cyclosporine treatment in a group of severe myasthenia gravis patients. *Journal of Neurology*, **1997**, 244, 542-7	5.5	51
447	LGMD2E patients risk developing dilated cardiomyopathy. *Neuromuscular Disorders*, **2003**, 13, 303-9	2.9	51
446	TNF-□nduced microRNAs Control Dystrophin Expression in Becker Muscular Dystrophy. *Cell Reports*, **2015**, 12, 1678-90	10.6	50
445	Molecular pathology and enzyme processing in various phenotypes of acid maltase deficiency. *Neurology*, **2008**, 70, 617-26	6.5	50

444	Coordinated and reversible reduction of enzymes involved in terminal oxidative metabolism in skeletal muscle mitochondria from a riboflavin-responsive, multiple acyl-CoA dehydrogenase deficiency patient. *Electrophoresis*, **2006**, 27, 1182-98	3.6	50
443	Prediction of myotonic dystrophy clinical severity based on the number of intragenic [CTG]n trinucleotide repeats. *American Journal of Medical Genetics Part A*, **1996**, 65, 342-7		50
442	The EuroBioBank Network: 10 years of hands-on experience of collaborative, transnational biobanking for rare diseases. *European Journal of Human Genetics*, **2015**, 23, 1116-23	5.3	49
441	Impaired autophagy contributes to muscle atrophy in glycogen storage disease type II patients. *Autophagy*, **2012**, 8, 1697-700	10.2	49
440	Right hemisphere dysfunction and emotional processing in ALS: an fMRI study. *Journal of Neurology* , **2010**, 257, 1970-8	5.5	49
439	Prevalence of dystrophin-positive fibers in 85 Duchenne muscular dystrophy patients. *Neuromuscular Disorders*, **1992**, 2, 41-5	2.9	49
438	Reliability of the El Escorial diagnostic criteria for amyotrophic lateral sclerosis. *Neuroepidemiology*, **2002**, 21, 265-70	5.4	48
437	Prognostic factors in mild dystrophinopathies. *Journal of the Neurological Sciences*, **1996**, 142, 70-8	3.2	48
436	Autophagy dysregulation in Danon disease. *Cell Death and Disease*, **2017**, 8, e2565	9.8	47
435	Radiological evidence of subclinical dysphagia in motor neuron disease. *Journal of Neurology*, **1998**, 245, 211-6	5.5	47
434	Clinical varieties of carnitine and carnitine palmitoyltransferase deficiency. *Clinical Biochemistry*, **1987**, 20, 1-7	3.5	47
433	Fingerprint body myopathy, a newly recognized congenital muscle disease. *Mayo Clinic Proceedings*, **1972**, 47, 377-88	6.4	47
432	Risk of arrhythmia in type I myotonic dystrophy: the role of clinical and genetic variables. *Journal of Neurology, Neurosurgery and Psychiatry*, **2009**, 80, 790-3	5.5	46
431	High plasma creatine kinase: review of the literature and proposal for a diagnostic algorithm. *Neurological Sciences*, **2006**, 27, 303-11	3.5	46
430	Targeting deregulated AMPK/mTORC1 pathways improves muscle function in myotonic dystrophy type I. *Journal of Clinical Investigation*, **2017**, 127, 549-563	15.9	45
429	Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study. *Orphanet Journal of Rare Diseases*, **2016**, 11, 34	4.2	45
428	Friedreich's disease: V. Variant form with vitamin E deficiency and normal fat absorption. *Neurology* , **1987**, 37, 68-74	6.5	45
427	Genotype-phenotype correlation in Pompe disease, a step forward. *Orphanet Journal of Rare Diseases*, **2014**, 9, 102	4.2	44

426	Protein and genetic diagnosis of limb girdle muscular dystrophy type 2A: The yield and the pitfalls. *Muscle and Nerve*, **2015**, 52, 163-73	3.4	44
425	Optimization of respiratory chain enzymatic assays in muscle for the diagnosis of mitochondrial disorders. *Mitochondrion*, **2011**, 11, 893-904	4.9	44
424	Myelo-optico-neuropathy in copper deficiency occurring after partial gastrectomy. Do small bowel bacterial overgrowth syndrome and occult zinc ingestion tip the balance?. *Journal of Neurology*, **2007**, 254, 1012-7	5.5	44
423	Clinical and molecular study in congenital muscular dystrophy with partial laminin alpha 2 (LAMA2) deficiency. *Human Mutation*, **2003**, 21, 103-11	4.7	44
422	Neuromuscular damage after hyperthermic isolated limb perfusion in patients with melanoma or sarcoma treated with chemotherapeutic agents. *Cancer Chemotherapy and Pharmacology*, **2000**, 46, 517-22	3.5	44
421	Correlation between clinical and molecular features in two MELAS families. *Journal of the Neurological Sciences*, **1992**, 113, 222-9	3.2	44
420	Carnitine deficiency: acute postpartum crisis. *Annals of Neurology*, **1978**, 4, 558-61	9.4	44
419	Clinical expression of facioscapulohumeral muscular dystrophy in carriers of 1-3 D4Z4 reduced alleles: experience of the FSHD Italian National Registry. *BMJ Open*, **2016**, 6, e007798	3	44
418	Molecular characterization of myophosphorylase deficiency in a group of patients from northern Italy. *Journal of the Neurological Sciences*, **1996**, 137, 14-9	3.2	43
417	Carnitine palmityl transferase deficiency: clinical variability, carrier detection, and autosomal-recessive inheritance. *Neurology*, **1981**, 31, 883-6	6.5	43
416	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. *JAMA Neurology*, **2018**, 75, 557-565	17.2	42
415	The clinical course of calpainopathy (LGMD2A) and dysferlinopathy (LGMD2B). *Neurological Research*, **2010**, 32, 41-6	2.7	42
414	The CTG repeat expansion size correlates with the splicing defects observed in muscles from myotonic dystrophy type 1 patients. *Journal of Medical Genetics*, **2008**, 45, 639-46	5.8	42
413	Calpain III mutation analysis of a heterogeneous limb-girdle muscular dystrophy population. *Neurology*, **1999**, 52, 1015-20	6.5	42
412	Genotype-phenotype correlations in a large series of patients with muscle type CPT II deficiency. *Neurological Research*, **2011**, 33, 24-32	2.7	41
411	Familial neuromuscular disease with tubular aggregates. *Muscle and Nerve*, **1985**, 8, 291-8	3.4	41
410	New motor outcome function measures in evaluation of late-onset Pompe disease before and after enzyme replacement therapy. *Muscle and Nerve*, **2012**, 45, 831-4	3.4	40
409	Clinical and molecular characterization of limb-girdle muscular dystrophy due to LAMA2 mutations. *Muscle and Nerve*, **2011**, 44, 703-9	3.4	40

408	Preferential central nucleation of type 2 myofibers is an invariable feature of myotonic dystrophy type 2. *Muscle and Nerve*, **2008**, 38, 1405-1411	3.4	40
407	Clinical-molecular correlation in 104 mild X-linked muscular dystrophy patients: characterization of sub-clinical phenotypes. *Neuromuscular Disorders*, **1994**, 4, 349-58	2.9	40
406	The natural history of cardiac involvement in myotonic dystrophy: an eight-year follow-up in 17 patients. *Clinical Cardiology*, **1988**, 11, 231-8	3.3	40
405	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. *PLoS ONE*, **2015**, 10, e0141240	3.7	40
404	LOPED study: looking for an early diagnosis in a late-onset Pompe disease high-risk population. *Journal of Neurology, Neurosurgery and Psychiatry*, **2016**, 87, 5-11	5.5	39
403	Calpain 3 is important for muscle regeneration: evidence from patients with limb girdle muscular dystrophies. *BMC Musculoskeletal Disorders*, **2012**, 13, 43	2.8	39
402	McArdle disease: the mutation spectrum of PYGM in a large Italian cohort. *Human Mutation*, **2006**, 27, 718	4.7	39
401	MYH7 gene mutation in myosin storage myopathy and scapulo-peroneal myopathy. *Neuromuscular Disorders*, **2007**, 17, 321-9	2.9	39
400	Cross-reactive protein in Duchenne muscle. *Lancet, The*, **1989**, 2, 1211-2	40	39
399	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. *Journal of Neurology*, **2016**, 263, 1204-14	5.5	39
398	Fatigue in muscular dystrophies. *Neuromuscular Disorders*, **2012**, 22 Suppl 3, S214-20	2.9	38
397	Cardiac transplantation in a Duchenne muscular dystrophy carrier. *Neuromuscular Disorders*, **1998**, 8, 585-90	2.9	38
396	Epidemiology of myotonic dystrophy in Italy: re-apprisal after genetic diagnosis. *Clinical Genetics*, **2001**, 59, 344-9	4	38
395	Epidemiology of spinal muscular atrophies in a sample of the Italian population. *Neuroepidemiology*, **1992**, 11, 34-8	5.4	38
394	Progress and challenges in diagnosis of dysferlinopathy. *Muscle and Nerve*, **2016**, 54, 821-835	3.4	38
393	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*, **2015**, 84, 1772-81	6.5	37
392	Relationship between neuropsychological impairment and grey and white matter changes in adult-onset myotonic dystrophy type 1. *NeuroImage: Clinical*, **2016**, 12, 190-7	5.3	37
391	Recessive MYL2 mutations cause infantile type I muscle fibre disease and cardiomyopathy. *Brain*, **2013**, 136, 282-93	11.2	37

390	Pilot trial of clenbuterol in spinal and bulbar muscular atrophy. *Neurology*, **2013**, 80, 2095-8	6.5	37
389	Muscle histopathology in myasthenia gravis with antibodies against MuSK and AChR. *Neuropathology and Applied Neurobiology*, **2009**, 35, 103-10	5.2	37
388	How to tackle the diagnosis of limb-girdle muscular dystrophy 2A. *European Journal of Human Genetics*, **2009**, 17, 598-603	5.3	37
387	Italian validation of INQoL, a quality of life questionnaire for adults with muscle diseases. *European Journal of Neurology*, **2010**, 17, 1178-1187	6	37
386	Risk prediction for clinical phenotype in myotonic dystrophy type 1: data from 2,650 patients. *Genetic Testing and Molecular Biomarkers*, **2007**, 11, 84-90		37
385	(CTG)n triplet mutation and phenotype manifestations in myotonic dystrophy patients. *Biochemical Medicine and Metabolic Biology*, **1993**, 50, 85-92		37
384	Plasma and urine carnitine levels during development. *Pediatric Research*, **1980**, 14, 1379	3.2	37
383	Enzyme replacement therapy for Pompe disease. *Current Neurology and Neuroscience Reports*, **2012** , 12, 70-5	6.6	36
382	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. *European Journal of Human Genetics*, **2015**, 23, 1254-8	5.3	36
381	Micro-RNAs in ALS muscle: Differences in gender, age at onset and disease duration. *Journal of the Neurological Sciences*, **2017**, 380, 58-63	3.2	35
380	Subacute sensory ataxia and optic neuropathy with thiamine deficiency. *Nature Reviews Neurology*, **2010**, 6, 288-93	15	35
379	Psychiatric disturbances associated with myasthenia gravis. *Acta Psychiatrica Scandinavica*, **1988**, 77, 443-5	6.5	35
378	Duchenne muscular dystrophy. A population study. *Human Genetics*, **1977**, 35, 225-31	6.3	35
377	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
376	Inhibition of proteasome activity promotes the correct localization of disease-causing alpha-sarcoglycan mutants in HEK-293 cells constitutively expressing beta-, gamma-, and delta-sarcoglycan. *American Journal of Pathology*, **2008**, 173, 170-81	5.8	34
375	Intronic breakpoint definition and transcription analysis in DMD/BMD patients with deletion/duplication at the 5Nmutation hot spot of the dystrophin gene. *Gene*, **2006**, 370, 26-33	3.8	34
374	Gene expression analysis in myotonic dystrophy: indications for a common molecular pathogenic pathway in DM1 and DM2. *Gene Expression*, **2007**, 13, 339-51	3.4	34
373	A novel laminin alpha2 isoform in severe laminin alpha2 deficient congenital muscular dystrophy. *Neurology*, **2000**, 55, 1128-34	6.5	34

372	Duchenne phenotype with in-frame deletion removing major portion of dystrophin rod: threshold effect for deletion size?. *Muscle and Nerve*, **1996**, 19, 1154-60	3.4	34
371	Dysferlinopathy course and sportive activity: clues for possible treatment. *Acta Myologica*, **2011**, 30, 127-32	1.6	34
370	Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPT-II deficiency. *Clinical Genetics*, **2012**, 82, 232-9	4	33
369	Facioscapulohumeral muscular dystrophy: a multicenter study on hearing function. *Audiology and Neuro-Otology*, **2008**, 13, 1-6	2.2	33
368	Dysferlin mutation analysis in a group of Italian patients with limb-girdle muscular dystrophy and Miyoshi myopathy. *European Journal of Neurology*, **2004**, 11, 657-61	6	33
367	Enormous dystrophin in a patient with Becker muscular dystrophy. *Neurology*, **1990**, 40, 808-12	6.5	33
366	"Mitochondrial neuropathies": A survey from the large cohort of the Italian Network. *Neuromuscular Disorders*, **2016**, 26, 272-6	2.9	32
365	Clinical and molecular cross-sectional study of a cohort of adult type III spinal muscular atrophy patients: clues from a biomarker study. *European Journal of Human Genetics*, **2013**, 21, 630-6	5.3	32
364	Myotonic dystrophy: tissue-specific effect of somatic CTG expansions on allele-specific DMAHP/SIX5 expression. *Human Molecular Genetics*, **1999**, 8, 1017-23	5.6	32
363	X-inactivation patterns in female Leber hereditary optic neuropathy patients do not support a strong X-linked determinant. *American Journal of Medical Genetics Part A*, **1996**, 61, 356-62		32
362	Male hypogonadism in myotonic dystrophy is related to (CTG)n triplet mutation. *Journal of Endocrinological Investigation*, **1994**, 17, 381-3	5.2	32
361	Clinical and biochemical aspects of carnitine deficiency and insufficiency: transport defects and inborn errors of beta-oxidation. *Critical Reviews in Clinical Laboratory Sciences*, **1992**, 29, 217-42	9.4	32
360	Population data on benign and severe forms of X-linked muscular dystrophy. *Human Genetics*, **1987**, 75, 217-20	6.3	32
359	Early ultrastructural and biochemical changes in muscle in dystrophia myotonica. *Journal of the Neurological Sciences*, **1970**, 10, 585-604	3.2	32
358	A new mutation in a family with cold-aggravated myotonia disrupts Na(+) channel inactivation. *Neurology*, **2001**, 56, 878-84	6.5	31
357	alpha-Sarcoglycan (adhalin) deficiency: complete deficiency patients are 5% of childhood-onset dystrophin-normal muscular dystrophy and most partial deficiency patients do not have gene mutations. *Journal of the Neurological Sciences*, **1996**, 140, 30-9	3.2	31
356	Novel missense mutations in PNPLA2 causing late onset and clinical heterogeneity of neutral lipid storage disease with myopathy in three siblings. *Molecular Genetics and Metabolism*, **2015**, 115, 110-7	3.7	29
355	Task force guidelines handbook: EFNS guidelines on diagnosis and management of fatty acid mitochondrial disorders. *European Journal of Neurology*, **2006**, 13, 923-9	6	29

354	Myoglobinuria and carnitine palmityltransferase (CPT) deficiency: studies with malonyl-CoA suggest absence of only CPT-II. *Neurology*, **1984**, 34, 353-6	6.5	29
353	Childhood encephalomyopathy with cytochrome c oxidase deficiency, ataxia, muscle wasting, and mental impairment. *Neurology*, **1986**, 36, 1048-52	6.5	29
352	Effects of combined endurance and resistance training in Amyotrophic Lateral Sclerosis: A pilot, randomized, controlled study. *European Journal of Translational Myology*, **2018**, 28, 7278	2.1	28
351	Telethon Network of Genetic Biobanks: a key service for diagnosis and research on rare diseases. *Orphanet Journal of Rare Diseases*, **2013**, 8, 129	4.2	28
350	Burden, professional support, and social network in families of children and young adults with muscular dystrophies. *Muscle and Nerve*, **2015**, 52, 13-21	3.4	28
349	Clinical and neuroimaging study of central nervous system in congenital myotonic dystrophy. *Journal of Neurology*, **1999**, 246, 186-92	5.5	28
348	Congenital muscular dystrophy: brain alterations in an unselected series of Western patients. *Journal of Neurology, Neurosurgery and Psychiatry*, **1991**, 54, 330-4	5.5	28
347	Subcellular distribution of acid and neutral alpha-glucosidases in normal, acid maltase deficient, and myophosphorylase deficient human skeletal muscle. *Archives of Biochemistry and Biophysics*, **1973**, 156, 350-5	4.1	28
346	Heterogeneous Phenotypes in Lipid Storage Myopathy Due to ETFDH Gene Mutations. *JIMD Reports*, **2018**, 38, 33-40	1.9	27
345	Skeletal muscle satellite cells in amyotrophic lateral sclerosis. *Ultrastructural Pathology*, **2014**, 38, 295-302	1.3	27
344	Muscle atrophy, ubiquitin-proteasome, and autophagic pathways in dysferlinopathy. *Muscle and Nerve*, **2014**, 50, 340-7	3.4	27
343	Myoclonus in mitochondrial disorders. *Movement Disorders*, **2014**, 29, 722-8	7	27
342	"I have got something positive out of this situation": psychological benefits of caregiving in relatives of young people with muscular dystrophy. *Journal of Neurology*, **2014**, 261, 188-95	5.5	27
341	Targeted array comparative genomic hybridization--a new diagnostic tool for the detection of large copy number variations in nemaline myopathy-causing genes. *Neuromuscular Disorders*, **2013**, 23, 56-65	2.9	27
340	The glucocorticoid receptor N363S polymorphism and steroid response in Duchenne dystrophy. *Journal of Neurology, Neurosurgery and Psychiatry*, **2006**, 77, 1177-9	5.5	27
339	Screening of calpain-3 autolytic activity in LGMD muscle: a functional map of CAPN3 gene mutations. *Journal of Medical Genetics*, **2007**, 44, 38-43	5.8	27
338	Congenital muscular dystrophy with partial merosin deficiency and late onset epilepsy. *European Neurology*, **1998**, 40, 37-45	2.1	27
337	L-carnitine uptake in differentiating human cultured muscle. *Biochimica Et Biophysica Acta - Molecular Cell Research*, **1991**, 1095, 217-22	4.9	27

336	Progress in Enzyme Replacement Therapy in Glycogen Storage Disease Type II. *Therapeutic Advances in Neurological Disorders*, **2009**, 2, 143-53	6.6	26
335	Emotional Lability in MND: Relationship to cognition and psychopathology and impact on caregivers. *Journal of the Neurological Sciences*, **2009**, 278, 16-20	3.2	26
334	Novel sarcoglycan gene mutations in a large cohort of Italian patients. *Journal of Medical Genetics*, **2003**, 40, e67	5.8	26
333	Heart transplantation in patients with inherited myopathies associated with end-stage cardiomyopathy: molecular and biochemical defects on cardiac and skeletal muscle. *Transplantation Proceedings*, **2001**, 33, 1596-9	1.1	26
332	Carnitine deficiency induced during haemodialysis. *Lancet, The*, **1978**, 1, 939	40	26
331	Spectrum of metabolic myopathies. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2015**, 1852, 615-21	6.9	25
330	Aberrant Compartment Formation by HSPB2 Mislocalizes Lamin A and Compromises Nuclear Integrity and Function. *Cell Reports*, **2017**, 20, 2100-2115	10.6	25
329	Natural history of upper motor neuron-dominant ALS. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2010**, 11, 424-9		25
328	Transcriptional and translational effects of intronic CAPN3 gene mutations. *Human Mutation*, **2010**, 31, E1658-69	4.7	25
327	Correlations between clinical severity, genotype and muscle pathology in limb girdle muscular dystrophy type 2A. *Journal of Medical Genetics*, **2007**, 44, 609-14	5.8	25
326	Esophageal motor function in patients with myotonic dystrophy. *Digestive Diseases and Sciences*, **1996**, 41, 2032-8	4	25
325	Impaired autophagy affects acid □glucosidase processing and enzyme replacement therapy efficacy in late-onset glycogen storage disease type II. *Neuropathology and Applied Neurobiology*, **2015**, 41, 672-5	5.2	24
324	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. *Scientific Reports*, **2016**, 6, 32439	4.9	24
323	Rippling muscle disease and facioscapulohumeral dystrophy-like phenotype in a patient carrying a heterozygous CAV3 T78M mutation and a D4Z4 partial deletion: Further evidence for "double trouble" overlapping syndromes. *Neuromuscular Disorders*, **2012**, 22, 534-40	2.9	24
322	Muscle atrophy in Limb Girdle Muscular Dystrophy 2A: a morphometric and molecular study. *Neuropathology and Applied Neurobiology*, **2013**, 39, 762-71	5.2	24
321	ALS risk but not phenotype is affected by ataxin-2 intermediate length polyglutamine expansion. *Neurology*, **2011**, 76, 2030-1	6.5	24
320	Decreased fatty acid beta-oxidation in riboflavin-responsive, multiple acylcoenzyme A dehydrogenase-deficient patients is associated with an increase in uncoupling protein-3. *Journal of Clinical Endocrinology and Metabolism*, **2003**, 88, 5921-6	5.6	24
319	Neonatal spinal muscular atrophy with diaphragmatic paralysis is unlinked to 5q11.2-q13. *Journal of Medical Genetics*, **1995**, 32, 216-9	5.8	24

318	Single muscle fibre analyses in 2 brothers with succinate dehydrogenase deficiency. *European Neurology*, **1994**, 34, 95-8	2.1	24
317	Review: Danon disease: Review of natural history and recent advances. *Neuropathology and Applied Neurobiology*, **2020**, 46, 303-322	5.2	24
316	Enzyme replacement therapy improves respiratory outcomes in patients with late-onset type II glycogenosis and high ventilator dependency. *Lung*, **2013**, 191, 537-44	2.9	23
315	Clinical phenotype, muscle MRI and muscle pathology of LGMD1F. *Journal of Neurology*, **2013**, 260, 2033-41	5.5	23
314	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. *Journal of Neurology*, **2017**, 264, 1777-1784	5.5	23
313	Fatigue and exercise intolerance in mitochondrial diseases. Literature revision and experience of the Italian Network of mitochondrial diseases. *Neuromuscular Disorders*, **2012**, 22 Suppl 3, S226-9	2.9	23
312	Cardiomyopathy in patients with POMT1-related congenital and limb-girdle muscular dystrophy. *European Journal of Human Genetics*, **2012**, 20, 1234-9	5.3	23
311	Muscle protein analysis in the detection of heterozygotes for recessive limb girdle muscular dystrophy type 2B and 2E. *Neuromuscular Disorders*, **2006**, 16, 792-9	2.9	23
310	Divergence of central nervous system involvement in 2 Italian sisters with congenital muscular dystrophy: a clinical and neuroradiological follow-up. *European Neurology*, **1995**, 35, 230-5	2.1	23
309	Clinical and hormonal aspects of male hypogonadism in myotonic dystrophy. *Italian Journal of Neurological Sciences*, **1996**, 17, 59-65		23
308	Characterization of two ETFDH mutations in a novel case of riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency. *Lipids in Health and Disease*, **2018**, 17, 254	4.4	23
307	An update on diagnostic options and considerations in limb-girdle dystrophies. *Expert Review of Neurotherapeutics*, **2018**, 18, 693-703	4.3	22
306	EFNS review on the role of muscle biopsy in the investigation of myalgia. *European Journal of Neurology*, **2013**, 20, 997-1005	6	22
305	Epidemiology of ALS in Padova district, Italy, from 1992 to 2005. *European Journal of Neurology*, **2009**, 16, 920-4	6	22
304	Activities of mitochondrial complexes correlate with nNOS amount in muscle from ALS patients. *Neuropathology and Applied Neurobiology*, **2007**, 33, 204-11	5.2	22
303	Myosin heavy chain composition of muscle fibers in spinal muscular atrophy. *Muscle and Nerve*, **1989**, 12, 43-51	3.4	22
302	Enzymes of the glycogen cycle and glycolysis in various human neuromuscular disorders. *Journal of Neurology, Neurosurgery and Psychiatry*, **1967**, 30, 411-5	5.5	22
301	Lipolysis and lipophagy in lipid storage myopathies. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2016**, 1862, 1367-73	6.9	22

300	208th ENMC International Workshop: Formation of a European Network to develop a European data sharing model and treatment guidelines for Pompe disease Naarden, The Netherlands, 26-28 September 2014. *Neuromuscular Disorders*, **2015**, 25, 674-8	2.9	21
299	Cognitive profile and MRI findings in limb-girdle muscular dystrophy 2I. *Journal of Neurology*, **2011**, 258, 1312-20	5.5	21
298	Steroids in muscular dystrophy: where do we stand?. *Neuromuscular Disorders*, **1998**, 8, 380-4	2.9	21
297	Amyotrophic lateral sclerosis with ragged-red fibers. *Archives of Neurology*, **2008**, 65, 403-6		21
296	LAMA2 loss-of-function mutation in a girl with a mild congenital muscular dystrophy. *Neurology*, **2004**, 63, 1118-21	6.5	21
295	Validity of hospital discharge diagnoses for the assessment of the prevalence and incidence of amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders: Official Publication of the World Federation of Neurology, Research Group on Motor Neuron Diseases*, **2001**, 2, 99-104		21
294	Prenatal diagnosis in congenital muscular dystrophy. *Lancet, The*, **1995**, 345, 591	40	21
293	Screening for mutations in the muscle promoter region and for exonic deletions in a series of 115 DMD and BMD patients. *Journal of Medical Genetics*, **1992**, 29, 127-30	5.8	21
292	Intellectual impairment and cognitive evoked potentials in myotonic dystrophy. *Journal of Nervous and Mental Disease*, **1989**, 177, 750-4	1.8	21
291	Adult acid maltase deficiency. Abnormalities in fibroblasts cultured from patients. *New England Journal of Medicine*, **1972**, 287, 948-51	59.2	21
290	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
289	Cerebral venous thrombosis at high altitude: A systematic review. *Revue Neurologique*, **2017**, 173, 189-193	3	20
288	TGFBR2 but not SPP1 genotype modulates osteopontin expression in Duchenne muscular dystrophy muscle. *Journal of Pathology*, **2012**, 228, 251-9	9.4	20
287	Psychopathological features and suicidal ideation in amyotrophic lateral sclerosis patients. *Neurological Sciences*, **2010**, 31, 735-40	3.5	20
286	Acetylcholinesterase activity is affected by stress conditions in Paracentrotus lividus coelomocytes. *Marine Biology*, **2003**, 143, 623-628	2.5	20
285	Cognitive and psychiatric evaluation of 40 patients with myotonic dystrophy. *Italian Journal of Neurological Sciences*, **1992**, 13, 53-8		20
284	Mitochondrial disorders of the nuclear genome. *Acta Myologica*, **2009**, 28, 16-23	1.6	20
283	Dominant muscular dystrophy with a novel SYNE1 gene mutation. *Muscle and Nerve*, **2015**, 51, 145-7	3.4	19

282	Alterations in Notch signalling in skeletal muscles from mdx and dko dystrophic mice and patients with Duchenne muscular dystrophy. *Experimental Physiology*, **2014**, 99, 675-87	2.4	19
281	Retrospective study on PET-SPECT imaging in a large cohort of myotonic dystrophy type 1 patients. *Neurological Sciences*, **2010**, 31, 757-63	3.5	19
280	Biochemical and ultrastructural evidence of endoplasmic reticulum stress in LGMD2I. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2007**, 451, 1047-55	5.1	19
279	Discordant clinical outcome in type III spinal muscular atrophy sibships showing the same deletion pattern. *Neuromuscular Disorders*, **1996**, 6, 261-4	2.9	19
278	Lipid storage myopathies. A review of metabolic defect and of treatment. *Journal of Neurology*, **1976**, 214, 1-11	5.5	19
277	Micro-RNA expression in muscle and fiber morphometry in myotonic dystrophy type 1. *Neurological Sciences*, **2017**, 38, 619-625	3.5	18
276	Diagnosis and management of autoimmune myasthenia gravis. *Clinical Drug Investigation*, **2011**, 31, 1-14	3.2	18
275	Comparison of muscle ultrastructure in myasthenia gravis with anti-MuSK and anti-AChR antibodies. *Journal of Neurology*, **2011**, 258, 746-52	5.5	18
274	Quality of life and motor impairment in ALS: Italian validation of ALSAQ. *Neurological Research*, **2010**, 32, 32-40	2.7	18
273	Sarcolemmal neuronal nitric oxide synthase defect in limb-girdle muscular dystrophy: an adverse modulating factor in the disease course?. *Journal of Neuropathology and Experimental Neurology*, **2009**, 68, 383-90	3.1	18
272	Large-scale disruption of microtubule pathways in morphologically normal human spastin muscle. *Neurology*, **2004**, 62, 1097-104	6.5	18
271	Detection of HTLV-I tax-rex and pol gene sequences of thymus gland in a large group of patients with myasthenia gravis. *Journal of Acquired Immune Deficiency Syndromes (1999)*, **2002**, 29, 300-6	3.1	18
270	Skeletal muscle sarcoplasmic reticulum phenotype in myotonic dystrophy. *Neuromuscular Disorders*, **1996**, 6, 33-47	2.9	18
269	Developmental patterns of LDH isozymes in fast and slow muscles of the rat. *Archives of Biochemistry and Biophysics*, **1970**, 141, 374-7	4.1	18
268	Limb-girdle muscular dystrophies: heterogeneity of clinical phenotypes and pathogenetic mechanisms. *Acta Myologica*, **2004**, 23, 130-6	1.6	18
267	Metabolic lipid muscle disorders: biomarkers and treatment. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419843359	6.6	17
266	Effects of short-to-long term enzyme replacement therapy (ERT) on skeletal muscle tissue in late onset Pompe disease (LOPD). *Neuropathology and Applied Neurobiology*, **2018**, 44, 449-462	5.2	17
265	Aberrant splicing and expression of the non muscle myosin heavy-chain gene MYH14 in DM1 muscle tissues. *Neurobiology of Disease*, **2012**, 45, 264-71	7.5	17

264	GYG1 gene mutations in a family with polyglucosan body myopathy. *Neurology: Genetics*, **2015**, 1, e21	3.8	17
263	Prevalence of asymptomatic vertebral fractures in late-onset Pompe disease. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 401-6	5.6	17
262	Two novel cosegregating mutations in tRNAMet and COX III, in a patient with exercise intolerance and autoimmune polyendocrinopathy. *Mitochondrion*, **2009**, 9, 123-9	4.9	17
261	Discordant clinical outcome in myotonic dystrophy relatives showing (CTG)n > 700 repeats. *Neuromuscular Disorders*, **1995**, 5, 157-9	2.9	17
260	Reappraisal of the incidence rate of Duchenne and Becker muscular dystrophies on the basis of molecular diagnosis. *Neuroepidemiology*, **1993**, 12, 326-30	5.4	17
259	Liver fatty acid-binding protein in two cases of human lipid storage. *Molecular and Cellular Biochemistry*, **1990**, 98, 225-30	4.2	17
258	Gender difference in limb-girdle muscular dystrophy: a muscle fiber morphometric study in 101 patients **2014**, 33, 179-85		17
257	Advances in imaging of brain abnormalities in neuromuscular disease. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419845567	6.6	16
256	Comparative transcriptional and biochemical studies in muscle of myotonic dystrophies (DM1 and DM2). *Neurological Sciences*, **2009**, 30, 185-92	3.5	16
255	Decreased expression of DMPK: correlation with CTG repeat expansion and fibre type composition in myotonic dystrophy type 1. *Neurological Sciences*, **2005**, 26, 235-42	3.5	16
254	Private beta- and gamma-sarcoglycan gene mutations: evidence of a founder effect in Northern Italy. *Human Mutation*, **2000**, 16, 13-7	4.7	16
253	Muscle glycerol kinase in Duchenne dystrophy and glycerol kinase deficiency. *Muscle and Nerve*, **1989**, 12, 307-13	3.4	16
252	Carnitine and acyltransferase in experimental neurogenic atrophies: changes with treatment. *Journal of Neurology*, **1984**, 231, 170-5	5.5	16
251	Muscle exercise in limb girdle muscular dystrophies: pitfall and advantages. *Acta Myologica*, **2015**, 34, 3-8	1.6	16
250	Longitudinal evaluation of SMN levels as biomarker for spinal muscular atrophy: results of a phase IIb double-blind study of salbutamol. *Journal of Medical Genetics*, **2019**, 56, 293-300	5.8	16
249	TDP-43 in skeletal muscle of patients affected with amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2010**, 11, 240-3		15
248	Metabolic myopathies: the challenge of new treatments. *Current Opinion in Pharmacology*, **2010**, 10, 338-45	5.1	15
247	Parkinson-like features in ALS with predominant upper motor neuron involvement. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2012**, 13, 137-43		15

246	A study on duplications of the dystrophin gene: evidence of a geographical difference in the distribution of breakpoints by intron. *Human Genetics*, **1994**, 94, 83-7	6.3	15
245	Effects of Functional Electrical Stimulation Lower Extremity Training in Myotonic Dystrophy Type I: A Pilot Controlled Study. *American Journal of Physical Medicine and Rehabilitation*, **2016**, 95, 809-817	2.6	15
244	MyomiRNAs Dysregulation in ALS Rehabilitation. *Brain Sciences*, **2019**, 9,	3.4	15
243	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. *JAMA Network Open*, **2020**, 3, e204040	10.4	14
242	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. *Neuromuscular Disorders*, **2018**, 28, 586-591	2.9	14
241	Genetic variation in KIFAP3 is associated with an upper motor neuron-predominant phenotype in amyotrophic lateral sclerosis. *Neurodegenerative Diseases*, **2011**, 8, 491-5	2.3	14
240	Multiple symmetric lipomatosis: evidence for mitochondrial dysfunction. *Journal of Clinical Neuromuscular Disease*, **2000**, 1, 124-30	1.1	14
239	Frequency of duplication at 17p11.2 in families of northeast Italy with Charcot-Marie-Tooth disease type 1. *Neuroepidemiology*, **1995**, 14, 49-53	5.4	14
238	Myophosphorylase deficiency affects muscle mitochondrial respiration as shown by 31P-MR spectroscopy in a case with associated multifocal encephalopathy. *Journal of the Neurological Sciences*, **1995**, 128, 84-91	3.2	14
237	Expression of muscle-type phosphorylase in innervated and aneural cultured muscle of patients with myophosphorylase deficiency. *Journal of Clinical Investigation*, **1993**, 92, 1774-80	15.9	14
236	Muscle fatigue, nNOS and muscle fiber atrophy in limb girdle muscular dystrophy. *Acta Myologica*, **2014**, 33, 119-26	1.6	14
235	Inhibition of muscle fibrosis results in increases in both utrophin levels and the number of revertant myofibers in Duchenne muscular dystrophy. *Oncotarget*, **2015**, 6, 23249-60	3.3	14
234	MicroRNAs and HDAC4 protein expression in the skeletal muscle of ALS patients **2020**, 39, 105-114		14
233	A mobile app for patients with Pompe disease and its possible clinical applications. *Neuromuscular Disorders*, **2018**, 28, 471-475	2.9	13
232	Update on polyglucosan storage diseases. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2019**, 475, 671-686	5.1	13
231	Ultrastructural changes in LGMD1F. *Neuropathology*, **2013**, 33, 276-80	2	13
230	Changes in skeletal muscle histology and metabolism in patients undergoing exercise deconditioning: effect of propionyl-L-carnitine. *Muscle and Nerve*, **1997**, 20, 1115-20	3.4	13
229	Cardioembolic stroke in Danon disease. *Clinical Genetics*, **2008**, 73, 388-90	4	13

228	Reduction of the DM-associated homeo domain protein (DMAHP) mRNA in different brain areas of myotonic dystrophy patients. *Neuromuscular Disorders*, **1999**, 9, 215-9	2.9	13
227	Hypertrophic cardiomyopathy with mitochondrial myopathy. A new phenotype of complex II defect. *International Heart Journal*, **1993**, 34, 63-77		13
226	Duchenne muscular dystrophy: data from family studies. *Human Genetics*, **1980**, 54, 63-8	6.3	13
225	Distinct disease phenotypes linked to different combinations of GAA mutations in a large late-onset GSDII sibship. *Orphanet Journal of Rare Diseases*, **2013**, 8, 159	4.2	12
224	Could utrophin rescue the myocardium of patients with dystrophin gene mutations?. *Journal of Molecular and Cellular Cardiology*, **1999**, 31, 1501-8	5.8	12
223	Postzygotic instability of the myotonic dystrophy p[AGC] in repeat supported by larger expansions in muscle and reduced amplifications in sperm. *Journal of Neurology*, **1995**, 242, 379-83	5.5	12
222	Letter: Duchenne carrier detection. *Lancet, The*, **1976**, 2, 90	40	12
221	Exercise-induced recurrent myoglobinuria: defective activity of inner carnitine palmitoyltransferase in muscle mitochondria of two patients. *Neurology*, **1987**, 37, 1184-8	6.5	12
220	Neuromuscular diseases and Covid-19: Advices from scientific societies and early observations in Italy. *European Journal of Translational Myology*, **2020**, 30, 9032	2.1	12
219	Muscle MRI in neutral lipid storage disease (NLSD). *Journal of Neurology*, **2017**, 264, 1334-1342	5.5	11
218	Hearing impairment in MELAS: new prospective in clinical use of microRNA, a systematic review. *Orphanet Journal of Rare Diseases*, **2018**, 13, 35	4.2	11
217	Improving the knowledge of amyotrophic lateral sclerosis genetics: novel SOD1 and FUS variants. *Neurobiology of Aging*, **2014**, 35, 1212.e7-1212.e10	5.6	11
216	An intronic mutation causes severe LGMD2A in a large inbred family belonging to a genetic isolate in the Alps. *Clinical Genetics*, **2012**, 82, 601-2	4	11
215	Abnormal expression of dysferlin in skeletal muscle and monocytes supports primary dysferlinopathy in patients with one mutated allele. *European Journal of Neurology*, **2011**, 18, 1021-3	6	11
214	Transcriptional behavior of DMD gene duplications in DMD/BMD males. *Human Mutation*, **2009**, 30, E31049	4.7	11
213	Positive correlation of CTG expansion and pharyngoesophageal alterations in myotonic dystrophy patients. *Italian Journal of Neurological Sciences*, **1998**, 19, 75-80		11
212	A pilot trial with clenbuterol in amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2006**, 7, 246-8		11
211	Novel spastin mutations and their expression analysis in two Italian families. *European Journal of Human Genetics*, **2003**, 11, 710-3	5.3	11

210	Regeneration in sarcoglycanopathies: expression studies of sarcoglycans and other muscle proteins. *Journal of the Neurological Sciences*, **1999**, 165, 170-7	3.2	11
209	Prevalent cardiac involvement in dystrophin Becker type mutation. *Neuromuscular Disorders*, **1994**, 4, 381-6	2.9	11
208	Diagnosis by protein analysis of dysferlinopathy in two patients mistaken as polymyositis. *Acta Myologica*, **2011**, 30, 185-7	1.6	11
207	Investigating the Mechanism of Chromosomal Deletion: Characterization of 39 Deletion Breakpoints in Introns 47 and 48 of the Human Dystrophin Gene **2002**, 80, 523-523		11
206	MiRNAs as biomarkers of phenotype in neutral lipid storage disease with myopathy. *Muscle and Nerve*, **2020**, 61, 253-257	3.4	11
205	Impaired copper and iron metabolism in blood cells and muscles of patients affected by copper deficiency myeloneuropathy. *Neuropathology and Applied Neurobiology*, **2014**, 40, 888-98	5.2	10
204	Muscle histopathology in upper motor neuron-dominant amyotrophic lateral sclerosis. *Amyotrophic Lateral Sclerosis and Other Motor Neuron Disorders*, **2008**, 9, 287-93		10
203	Association between human polymorphic DNA markers and hypoxia adaptation in Sherpa detected by a preliminary genome scan. *Annals of Human Genetics*, **2007**, 71, 630-8	2.2	10
202	A truncation in the RYR1 gene associated with central core lesions in skeletal muscle fibres. *Journal of Medical Genetics*, **2007**, 44, e67	5.8	10
201	Acute quadriplegic myopathy in a 17-month-old boy. *Journal of Child Neurology*, **2000**, 15, 63-6	2.5	10
200	Motor neuron disease in the Padua district of Italy: an epidemiological study. *Neuroepidemiology*, **1996**, 15, 173-9	5.4	10
199	Residual muscle cytochrome c oxidase activity accounts for submaximal exercise lactate threshold in chronic progressive external ophthalmoplegia. *Muscle and Nerve*, **1996**, 19, 342-9	3.4	10
198	Next generation sequencing detection of late onset pompe disease. *Muscle and Nerve*, **2016**, 53, 981-3	3.4	10
197	Identification of an intragenic deletion in the SGCB gene through a re-evaluation of negative next generation sequencing results. *Neuromuscular Disorders*, **2016**, 26, 367-9	2.9	10
196	Pathogenesis, clinical features and diagnosis of sarcoglycanopathies. *Expert Opinion on Orphan Drugs*, **2016**, 4, 1239-1251	1.1	9
195	Familial polyglucosan body myopathy with unusual phenotype. *Neuropathology and Applied Neurobiology*, **2015**, 41, 385-90	5.2	9
194	Incomplete penetrance in limb-girdle muscular dystrophy type 1F. *Muscle and Nerve*, **2015**, 52, 305-6	3.4	9
193	Autonomic regulation in muscular dystrophy. *Frontiers in Physiology*, **2013**, 4, 257	4.6	9

192	Prevalence of unsuspected myopathy in infants presenting for clubfoot surgery. *Paediatric Anaesthesia*, **1995**, 5, 165-70	1.8	9
191	Letter: Carnitine deficiency. *Lancet, The*, **1975**, 2, 554	40	9
190	A 5-year clinical follow-up study from the Italian National Registry for FSHD. *Journal of Neurology*, **2021**, 268, 356-366	5.5	9
189	Elevated Expression of Moesin in Muscular Dystrophies. *American Journal of Pathology*, **2017**, 187, 654-668	3.8	8
188	MyomiRNAs and myostatin as physical rehabilitation biomarkers for myotonic dystrophy. *Neurological Sciences*, **2020**, 41, 2953-2960	3.5	8
187	New treatments for myasthenia: a focus on antisense oligonucleotides. *Drug Design, Development and Therapy*, **2013**, 7, 13-7	4.4	8
186	Long-term follow-up results in enzyme replacement therapy for Pompe disease: a case report. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S389-93	5.4	8
185	Late-onset GSDII with novel GAA gene mutation. *Clinical Genetics*, **2007**, 71, 374-5	4	8
184	Co-segregation of LMNA and PMP22 gene mutations in the same family. *Neuromuscular Disorders*, **2005**, 15, 858-62	2.9	8
183	Infantile lipid storage myopathy with nocturnal hypoventilation shows abnormal low-affinity muscle carnitine uptake in vitro. *Neuromuscular Disorders*, **1999**, 9, 320-2	2.9	8
182	Non-radioactive detection of 17p11.2 duplication in CMT1A: a study of 78 patients. *Journal of Medical Genetics*, **1994**, 31, 880-3	5.8	8
181	Genetic epidemiology of myotonic dystrophy. *Genetic Epidemiology*, **1987**, 4, 289-98	2.6	8
180	Current and emerging therapies in Becker muscular dystrophy (BMD). *Acta Myologica*, **2019**, 38, 172-179	1.6	8
179	LGMD. Identification, description and classification. *Acta Myologica*, **2020**, 39, 207-217	1.6	8
178	Neuromuscular diseases and Covid-19: Advices from scientific societies and early observations in Italy. *European Journal of Translational Myology*, **2020**, 30, 286-290	2.1	8
177	Large genotype-phenotype study in carriers of D4Z4 borderline alleles provides guidance for facioscapulohumeral muscular dystrophy diagnosis. *Scientific Reports*, **2020**, 10, 21648	4.9	8
176	CAG repeat length in androgen receptor gene is not associated with amyotrophic lateral sclerosis. *European Journal of Neurology*, **2012**, 19, 1373-5	6	7
175	Sphingomonas paucimobilis associated with localised calf myositis. *Journal of Neurology, Neurosurgery and Psychiatry*, **2008**, 79, 1194-5	5.5	7

174	Hereditary spastic paraplegia associated with peripheral neuropathy: a distinct clinical and genetic entity. *Neuromuscular Disorders*, **2000**, 10, 497-502	2.9	7
173	Occurrence of two different intragenic deletions in two male relatives affected with Duchenne muscular dystrophy. *American Journal of Medical Genetics Part A*, **1994**, 50, 84-6		7
172	Absence of dystrophin and spectrin in regenerating muscle fibers from Becker dystrophy patients. *Journal of the Neurological Sciences*, **1994**, 123, 88-94	3.2	7
171	The role of life events in the myasthenia gravis outcome: a one-year longitudinal study. *Acta Neurologica Scandinavica*, **1989**, 79, 288-91	3.8	7
170	Defects of fatty-acid oxidation in muscle. *Baillieres Clinical Endocrinology and Metabolism*, **1990**, 4, 561-82		7
169	Malonyl-CoA abnormal inhibition of residual enzyme activity in carnitine palmitoyltransferase deficiency. *European Neurology*, **1986**, 25, 309-16	2.1	7
168	Undiagnosed myopathy before surgery and safe anaesthesia table. *Acta Myologica*, **2013**, 32, 100-5	1.6	7
167	ETF dehydrogenase advances in molecular genetics and impact on treatment. *Critical Reviews in Biochemistry and Molecular Biology*, **2021**, 56, 360-372	8.7	7
166	Clinical and genetic characterization of an Italian family with slow-channel syndrome. *Neurological Sciences*, **2019**, 40, 503-507	3.5	7
165	Limb girdle muscular dystrophies: clinical-genetical diagnostic update and prospects for therapy. *Expert Opinion on Orphan Drugs*, **2017**, 5, 769-784	1.1	6
164	Hereditary protein C deficiency associated with riboflavin-responsive lipid storage myopathy. *European Journal of Neurology*, **1996**, 3, 61-65	6	6
163	Autoimmune neuromuscular disease induced by a preparation of choline acetyltransferase. *Experimental Neurology*, **1982**, 75, 23-35	5.7	6
162	Geographic distribution of hereditary myopathies in northeast Italy. *Social Biology*, **1974**, 21, 235-41		6
161	Relationship of serum enzyme changes to muscle damage in vitamin E deficiency of the rabbit. *Lo Sperimentale*, **1968**, 118, 349-69		6
160	Microglia polarization by mitochondrial metabolism modulation: A therapeutic opportunity in neurodegenerative diseases. *Mitochondrion*, **2019**, 46, 334-336	4.9	6
159	Circulating miR-206 as a Biomarker for Patients Affected by Severe Limb Girdle Muscle Dystrophies. *Genes*, **2021**, 12,	4.2	6
158	Transportin 3 (TNPO3) and related proteins in limb girdle muscular dystrophy D2 muscle biopsies: A morphological study and pathogenetic hypothesis. *Neuromuscular Disorders*, **2020**, 30, 685-692	2.9	5
157	Correlation between ETFDH mutations and dysregulation of serum myomiRs in MADD patients. *European Journal of Translational Myology*, **2020**, 30, 8880	2.1	5

156	A new family with transportinopathy: increased clinical heterogeneity. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419850433	6.6	5
155	Therapeutic advances in the management of Pompe disease and other metabolic myopathies. *Therapeutic Advances in Neurological Disorders*, **2013**, 6, 311-21	6.6	5
154	A novel out-of-frame mutation in the neurofilament light chain gene (NEFL) does not result in Charcot-Marie-Tooth disease type 2E. *Neurogenetics*, **2005**, 6, 49-50	3	5
153	Serum lipids, lipoprotein analysis and apoprotein A-I, A-II and B levels in Friedreich's ataxia. *European Neurology*, **1990**, 30, 132-7	2.1	5
152	Evolution of cardiac involvement in progressive ophthalmoplegia with deleted mitochondrial DNA. *International Heart Journal*, **1990**, 31, 115-20		5
151	Myopathological findings in progressive myoclonus epilepsy. *Acta Neuropathologica Supplementum*, **1981**, 7, 334-7		5
150	MiRNAs, Myostatin, and Muscle MRI Imaging as Biomarkers of Clinical Features in Becker Muscular Dystrophy. *Diagnostics*, **2020**, 10,	3.8	5
149	Generation of induced Pluripotent Stem Cells as disease modelling of NLSDM. *Molecular Genetics and Metabolism*, **2017**, 121, 28-34	3.7	4
148	Interpretation of the Epigenetic Signature of Facioscapulohumeral Muscular Dystrophy in Light of Genotype-Phenotype Studies. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	4
147	Neuromuscular disease. Diagnosis and discovery in limb-girdle muscular dystrophy. *Nature Reviews Neurology*, **2016**, 12, 6-8	15	4
146	Genetic Neuromuscular Disorders **2014**,		4
145	Prevention of cardiomyopathy in Duchenne muscular dystrophy. *Lancet Neurology, The*, **2015**, 14, 127-8	24.1	4
144	Autophagy in Natural History and After ERT in Glycogenosis Type II. *JIMD Reports*, **2015**, 21, 71-7	1.9	4
143	The role of ultrastructural examination in storage diseases. *Ultrastructural Pathology*, **2010**, 34, 243-51	1.3	4
142	Novel mutations and polymorphisms in the human dystrophin gene detected by double-strand conformation analysis. *Human Mutation*, **1997**, 9, 188-90	4.7	4
141	Childhood dermatomyositis associated with intracranial tumor and liver cysts. *European Journal of Paediatric Neurology*, **2007**, 11, 76-80	3.8	4
140	Expression profiling characterization of laminin alpha-2 positive MDC. *Biochemical and Biophysical Research Communications*, **2006**, 350, 345-51	3.4	4
139	Correlation of clinical function and muscle CT scan images in limb-girdle muscular dystrophy. *Neurological Sciences*, **2000**, 21, S975-7	3.5	4

138	Dystrophin-positive myotubes in innervated muscle cultures from Duchenne and Becker muscular dystrophy patients. *Neuromuscular Disorders*, **1993**, 3, 119-27	2.9	4
137	Variability of the expression of muscle mitochondrial damage in ocular mitochondrial myopathy. *Neuromuscular Disorders*, **1992**, 2, 397-404	2.9	4
136	Further evidence of a duplication in 17p11.2 in families with recurrence of HMSN Ia (Charcot-Marie-Tooth neuropathy type Ia). *Human Genetics*, **1992**, 90, 231-4	6.3	4
135	Assessment of the value of thymic scan in myasthenia gravis. *Journal of Neurology*, **1979**, 220, 21-9	5.5	4
134	Abnormal myomuscular junctions and AChE in a congenital neuromuscular disease. *Muscle and Nerve*, **1980**, 3, 240-7	3.4	4
133	Guanidine hydrochloride in infantile and juvenile spinal muscular atrophy. A double blind controlled study. *Acta Neurologica*, **1980**, 2, 460-5		4
132	Central Nervous System Involvement as Outcome Measure for Clinical Trials Efficacy in Myotonic Dystrophy Type 1. *Frontiers in Neurology*, **2020**, 11, 624	4.1	4
131	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. *Genes*, **2018**, 9,	4.2	4
130	Morphological study of TNPO3 and SRSF1 interaction during myogenesis by combining confocal, structured illumination and electron microscopy analysis. *Molecular and Cellular Biochemistry*, **2021**, 476, 1797-1811	4.2	4
129	Morphological changes in late onset acid Maltase deficient patients with splicing gene mutation. *Acta Myologica*, **2003**, 22, 90-6	1.6	4
128	ATP1A3 mutant patient with alternating hemiplegia of childhood and brain spectroscopic abnormalities. *Journal of the Neurological Sciences*, **2017**, 379, 36-38	3.2	3
127	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, 414-22	4.2	3
126	Neuromuscular diseases: advances in therapy and diagnosis. *Lancet Neurology, The*, **2012**, 11, 15-7	24.1	3
125	P.6.4 Salbutamol tolerability and efficacy in adult type III SMA patients: Results of a multicentric, molecular and clinical, double-blind, placebo-controlled study. *Neuromuscular Disorders*, **2013**, 23, 771	2.9	3
124	Disorders of lipid metabolism. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2007**, 86, 183-91	3	3
123	G.P.4.07 Relation between LGMD2B progression and physical activity. *Neuromuscular Disorders*, **2007**, 17, 789	2.9	3
122	Prenatal diagnosis in a family affected with beta-sarcoglycan muscular dystrophy. *Neuromuscular Disorders*, **1999**, 9, 323-5	2.9	3
121	Glucose and ketone body turnover in carnitine-palmitoyl-transferase deficiency. *Metabolism: Clinical and Experimental*, **1987**, 36, 821-6	12.7	3

120	State of the art in muscle glycogenoses. *Acta Myologica*, **2010**, 29, 339-42	1.6	3
119	A multicenter, double-blind, randomized trial of deflazacort versus prednisone in Duchenne muscular dystrophy **2000**, 23, 1344		3
118	The clinical and molecular spectrum of autosomal dominant limb-girdle muscular dystrophies focusing on transportinopathy. *Expert Opinion on Orphan Drugs*, **2019**, 7, 223-232	1.1	2
117	G.P.251. *Neuromuscular Disorders*, **2014**, 24, 892	2.9	2
116	Drugs in development and dietary approach for Duchenne muscular dystrophy. *Orphan Drugs: Research and Reviews*, **2015**, 51		2
115	Chapter 31: muscular dystrophy. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2010**, 95, 477-88	3	2
114	Increased aldosterone levels in acute mountain sickness at Capanna Regina Margherita. *Wilderness and Environmental Medicine*, **1997**, 8, 247-9	1.4	2
113	Defective assembly of sarcoglycan complex in patients with beta-sarcoglycan gene mutations. Study of aneural and innervated cultured myotubes. *Neuropathology and Applied Neurobiology*, **2002**, 28, 190-9	5.2	2
112	New therapies in muscular dystrophies. *Neurological Sciences*, **2000**, 21, S919-24	3.5	2
111	Audit of care of acute inflammatory polyradiculoneuropathy in Italy. *Pharmacoepidemiology and Drug Safety*, **1995**, 4, 137-146	2.6	2
110	Toxic myopathy induced by industrial minerals oils: clinical and histopathological features. *Italian Journal of Neurological Sciences*, **1986**, 7, 599-604		2
109	Myotonic dystrophy and chromosome translocation segregating in the same family. *Journal of Neurogenetics*, **1987**, 4, 47-56	1.6	2
108	Peroneal muscular atrophy with ataxia and partial myoclonic epilepsy. *Journal of Neurology*, **1981**, 226, 1-13	5.5	2
107	Duchenne carrier detection. *Lancet, The*, **1976**, 2, 415-6	40	2
106	Heterogeneity of Charcot-Marie-Tooth disease suggested by a linkage study. *Advances in Neurology* , **1988**, 48, 209-19		2
105	Challenges and progress in the diagnosis of Congenital Muscular Dystrophies. *Expert Opinion on Orphan Drugs*, **2016**, 4, 347-358	1.1	2
104	Fatty Acid Mitochondrial Disorders501-511		2
103	Familial ALS: clinical, genetic and morphological features. *Advances in Experimental Medicine and Biology*, **1987**, 209, 109-10	3.6	2

102	The role of transmission electron microscopy in vacuole-associated myopathies. *Ultrastructural Pathology*, **2017**, 41, 88-90	1.3	1
101	Can miR-34a be suitable for monitoring sensorineural hearing loss in patients with mitochondrial disease? A case series. *International Journal of Neuroscience*, **2020**, 130, 1272-1277	2	1
100	Duchenne Muscular Dystrophy **2018**, 3-7		1
99	Mitochondrial Complex I Deficiency due to ACAD9 Deficiency **2018**, 291-292		1
98	Enzyme replacement therapy for the treatment of Pompe disease. *Expert Opinion on Orphan Drugs*, **2018**, 6, 311-318	1.1	1
97	New Pathogenetic Mechanisms that Link Autophagy to Pompe Disease. *Journal of Neuromuscular Diseases*, **2015**, 2, S9-S9	5	1
96	Multifactorial study of inflammatory myopathies. Report of 29 cases. *Italian Journal of Neurological Sciences*, **1993**, 14, 69-76		1
95	Fatal lipid storage with abnormal mitochondria in an infant. *Acta Neuropathologica Supplementum*, **1981**, 7, 221-5		1
94	HEREDITARY SPHEROCYTOSIS IN PERONEAL MUSCULAR ATROPHY **1978**, 435-440		1
93	Assessing diagnosis and managing respiratory and cardiac complications of sarcoglycanopathy. *Expert Opinion on Orphan Drugs*, **2020**, 8, 515-523	1.1	1
92	Epidemiology of motor neuron disease in north-east Veneto region: Venice, Padua, and Belluno Provinces (1972-1981). *Advances in Experimental Medicine and Biology*, **1987**, 209, 333-7	3.6	1
91	European Federation of the Neurological Societies guidelines on the diagnostic approach to paucisymptomatic or asymptomatic hyperCKemia. *Muscle and Nerve*, **2020**, 61, E14-E15	3.4	1
90	Diagnostic challenges in metabolic myopathies. *Expert Review of Neurotherapeutics*, **2020**, 20, 1287-1298	4.3	1
89	Exercise, nutrition and enzyme replacement therapy are efficacious in adult Pompe patients: report from EPOC Consortium. *European Journal of Translational Myology*, **2021**, 31,	2.1	1
88	Recurrent N209* ABHD5 mutation in two unreported families with Chanarin Dorfman Syndrome. *European Journal of Translational Myology*, **2021**, 31,	2.1	1
87	MicroRNAs are appropriate in mitochondrial related hearing loss? Answer to the skepticism. *Orphanet Journal of Rare Diseases*, **2018**, 13, 119	4.2	1
86	An updated review on the role of prescribed exercise in the management of Amyotrophic lateral sclerosis. *Expert Review of Neurotherapeutics*, **2021**, 21, 871-879	4.3	1
85	Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. *Journal of Neuromuscular Diseases*, **2015**, 2, S36-S37	5	0

84	Cardiomyopathy in a patient with limb-girdle muscular dystrophy type 2D: Pathomorphological aspects. *Forensic Science International: Genetics Supplement Series*, **2009**, 1, 58-62		0
83	Recommendations for traveling to altitude with neurological disorders.. *Journal of Central Nervous System Disease*, **2021**, 13, 11795735211053448	4.4	0
82	Limb-Girdle Muscular Dystrophy Type 1F **2018**, 41-44		
81	Limb-Girdle Muscular Dystrophy Type 2A **2018**, 45-50		
80	Limb-Girdle Muscular Dystrophy Type 2B **2018**, 51-55		
79	Limb-Girdle Muscular Dystrophy Type 2C **2018**, 57-60		
78	Limb-Girdle Muscular Dystrophy Type 2D **2018**, 61-64		
77	Limb-Girdle Muscular Dystrophy Type 2E **2018**, 65-69		
76	Limb-Girdle Muscular Dystrophy Type 2H **2018**, 73-74		
75	Limb-Girdle Muscular Dystrophy Type 2I **2018**, 75-78		
74	Limb-Girdle Muscular Dystrophy Type 2J **2018**, 79-81		
73	Duchenne Muscular Dystrophy Carrier **2018**, 9-12		
72	Limb-Girdle Muscular Dystrophy Type 2L **2018**, 87-89		
71	Limb-Girdle Muscular Dystrophy Type 2T **2018**, 97-98		
70	Congenital Muscular Dystrophy Type 1A **2018**, 99-103		
69	Fukuyama Congenital Muscular Dystrophy: Walker-Warburg Syndrome **2018**, 107-110		
68	Becker Muscular Dystrophy **2018**, 13-16		
67	Oculopharyngeal Muscular Dystrophy **2018**, 133-134		

66 Emery-Dreifuss Muscular Dystrophy Type 4 **2018**, 25-28

65 Limb-Girdle Muscular Dystrophy Type 1A **2018**, 29-31

64 Limb-Girdle Muscular Dystrophy Type 1C **2018**, 37-40

63 Congenital Nemaline Myopathy Type 1 **2018**, 159-160

62 Congenital Nemaline Myopathy Type 2 **2018**, 161-165

61 Congenital Myofibrillar Myopathy Type 2 **2018**, 179-180

60 Congenital Myofibrillar Myopathy Type 5 **2018**, 181-182

59 Vacuolar Myopathy with Calsequestrin Aggregates **2018**, 191-193

58 Myotonic Dystrophy Type 2, Proximal Myotonic Myopathy **2018**, 209-212

57 Hypokalemic Periodic Paralysis Type 1 **2018**, 221-222

56 Congenital Myasthenic Syndrome Type 11 **2018**, 233-234

55 Glycogenosis Type 2, Pompe Disease **2018**, 241-247

54 Glycogenosis Type 4, Andersen Disease **2018**, 253-254

53 Glycogenosis Type 5, McArdle Disease **2018**, 255-257

52 Glycogenosis Type 7, Tarui Disease **2018**, 259-260

51 Polyglucosan Body Myopathy Type 2 **2018**, 265-268

50 Chronic Progressive External Ophthalmoplegia **2018**, 273-276

49 MELAS (Myopathy, Encephalopathy, Lactic Acidosis, Stroke-Like Episodes) **2018**, 277-282

48 Mitochondrial Encephalomyopathy with COX Deficiency **2018**, 287-290

47 Spastic Paraparesis Type 4 **2018**, 403-405

46 Optic Atrophy Plus Syndrome **2018**, 411-413

45 Amyotrophic Lateral Sclerosis Type 2, Juvenile **2018**, 419-421

44 Systemic Primary Carnitine Deficiency **2018**, 307-311

43 Spinal Muscular Atrophy Type 1, Werdnig-Hoffmann Disease **2018**, 341-343

42 Spinal Muscular Atrophy Type 2, Werdnig-Hoffmann Disease **2018**, 345-347

41 Spinal Muscular Atrophy Type 3, Kugelberg-Welander Disease **2018**, 349-351

40 Spinal Bulbar Muscular Atrophy, Kennedy Disease **2018**, 353-356

39 Charcot-Marie-Tooth Neuropathy with Pyramidal Features **2018**, 375-378

38 Distal Hereditary Motor Neuropathy Type 2C **2018**, 389-390

37 Limb-Girdle Muscular Dystrophy with Epidermolysis Bullosa Simplex **2018**, 93-95

36 Myotonic Dystrophy Type 1, Steinert Disease **2018**, 199-203

35 Facioscapulohumeral Muscular Dystrophy Type 1A **2018**, 123-127

34 Endocrinological Myopathies **2016**, 143-154

33 Response to: Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. *Neuromuscular Disorders*, **2016**, 26, 549 2.9

32 Becker Muscular Dystrophy **2014**, 13-17

31 Facioscapulohumeral Muscular Dystrophy Type 1A **2014**, 105-109

30	Duchenne Muscular Dystrophy **2014**, 3-7	
29	Pompe Disease Diagnosis, Treatment, and Outcomes in Italy: Pompe Disease Registry Data from Italy Compared with the Rest-of-World. *Clinical Therapeutics*, **2011**, 33, S35	3.5
28	Nutritional Recommendations for Patients with Glycogen Storage Disease Type II. *Clinical Therapeutics*, **2008**, 30, S21	3.5
27	Fatty Acid Mitochondrial Disorders526-533	
26	cDNA Sequencing of Nuclear NADH Dehydrogenase Subunit Genes in Complex I Deficient Myopathic Patients. *Gene Function & Disease*, **2000**, 1, 21-27	
25	European consensus for starting and stopping enzyme replacement therapy in adult patients with Pompe disease: a 10-year experience. *Nervno-Myshechnye Bolezni*, **2019**, 8, 19-34	0.2
24	Various clinical presentation of mitochondriopathies: clinical and therapeutic considerations **1991**, 255-262	
23	Diagnostic Approach to Pauci- or Asymptomatic hyperCKemia279-286	
22	Duchenne Muscular Dystrophy Carrier **2014**, 9-12	
21	Limb-Girdle Muscular Dystrophy Type 2E **2014**, 61-65	
20	Glycogenosis Type 5, McArdle Disease **2014**, 217-219	
19	Chronic Progressive External Ophthalmoplegia **2014**, 229-232	
18	Congenital Nemaline Myopathy Type 2 **2014**, 137-140	
17	Limb-Girdle Muscular Dystrophy Type 2I **2014**, 71-74	
16	Mitochondrial Encephalomyopathy with COX Deficiency **2014**, 243-246	
15	Limb-Girdle Muscular Dystrophy Type 1C **2014**, 31-34	
14	Myotonic Dystrophy Type 2, Proximal Myotonic Myopathy **2014**, 177-180	
13	Spinal Muscular Atrophy Type 1, Werdnig-Hoffmann Disease **2014**, 295-297	

List of Publications

12	Systemic Primary Carnitine Deficiency **2014**, 261-265	
11	Limb-Girdle Muscular Dystrophy Type 1F **2014**, 35-38	
10	Limb-Girdle Muscular Dystrophy Type 2B **2014**, 47-52	
9	Spinal Muscular Atrophy Type 3, Kugelberg-Welander Disease **2014**, 303-305	
8	Congenital Muscular Dystrophy Type 1A **2014**, 83-87	
7	Glycogenosis Type 2, Pompe Disease **2014**, 205-211	
6	Travel to altitude with neurological disorders recommendation of the UIAA Medical Commission. *Health Promotion & Physical Activity*, **2021**, 15, 29-39	0.1
5	Reply. *Muscle and Nerve*, **2016**, 53, 157-8	3.4
4	Authors reply. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419878316	6.6
3	Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. *Journal of Neuromuscular Diseases*, **2015**, 2, S36-S37	5
2	LGMD D2 TNPO3-Related: From Clinical Spectrum to Pathogenetic Mechanism.. *Frontiers in Neurology*, **2022**, 13, 840683	4.1
1	Distinct Phenotypic and microRNA Expression in X-Linked Charcot Marie Tooth Correlated with a Novel Mutation in the GJB1 Gene **2022**, 1, 66-74	