Corrado Angelini

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	LGMD D2 TNPO3-Related: From Clinical Spectrum to Pathogenetic Mechanism.. *Frontiers in Neurology*, **2022**, 13, 840683	4.1	
568	Distinct Phenotypic and microRNA Expression in X-Linked Charcot Marie Tooth Correlated with a Novel Mutation in the GJB1 Gene **2022**, 1, 66-74		
567	Recommendations for traveling to altitude with neurological disorders.. *Journal of Central Nervous System Disease*, **2021**, 13, 11795735211053448	4.4	0
566	ETF dehydrogenase advances in molecular genetics and impact on treatment. *Critical Reviews in Biochemistry and Molecular Biology*, **2021**, 56, 360-372	8.7	7
565	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
564	Recurrent N209* ABHD5 mutation in two unreported families with Chanarin Dorfman Syndrome. *European Journal of Translational Myology*, **2021**, 31,	2.1	1
563	Travel to altitude with neurological disorders Recommendation of the UIAA Medical Commission. *Health Promotion & Physical Activity*, **2021**, 15, 29-39	0.1	
562	A 5-year clinical follow-up study from the Italian National Registry for FSHD. *Journal of Neurology*, **2021**, 268, 356-366	5.5	9
561	Circulating miR-206 as a Biomarker for Patients Affected by Severe Limb Girdle Muscle Dystrophies. *Genes*, **2021**, 12,	4.2	6
560	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
559	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
558	Phenotypic Variability Among Patients With D4Z4 Reduced Allele Facioscapulohumeral Muscular Dystrophy. *JAMA Network Open*, **2020**, 3, e204040	10.4	14
557	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
556	Correlation between ETFDH mutations and dysregulation of serum myomiRs in MADD patients. *European Journal of Translational Myology*, **2020**, 30, 8880	2.1	5
555	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
554	MyomiRNAs and myostatin as physical rehabilitation biomarkers for myotonic dystrophy. *Neurological Sciences*, **2020**, 41, 2953-2960	3.5	8
553	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

552	LGMD. Identification, description and classification. *Acta Myologica*, **2020**, 39, 207-217	1.6	8
551	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
550	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
549	MicroRNAs and HDAC4 protein expression in the skeletal muscle of ALS patients **2020**, 39, 105-114		14
548	Assessing diagnosis and managing respiratory and cardiac complications of sarcoglycanopathy. *Expert Opinion on Orphan Drugs*, **2020**, 8, 515-523	1.1	1
547	Review: Danon disease: Review of natural history and recent advances. *Neuropathology and Applied Neurobiology*, **2020**, 46, 303-322	5.2	24
546	MiRNAs as biomarkers of phenotype in neutral lipid storage disease with myopathy. *Muscle and Nerve*, **2020**, 61, 253-257	3.4	11
545	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
544	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
543	Diagnostic challenges in metabolic myopathies. *Expert Review of Neurotherapeutics*, **2020**, 20, 1287-1298	4.3	1
542	MiRNAs, Myostatin, and Muscle MRI Imaging as Biomarkers of Clinical Features in Becker Muscular Dystrophy. *Diagnostics*, **2020**, 10,	3.8	5
541	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
540	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
539	Advances in imaging of brain abnormalities in neuromuscular disease. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419845567	6.6	16
538	Metabolic lipid muscle disorders: biomarkers and treatment. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419843359	6.6	17
537	A new family with transportinopathy: increased clinical heterogeneity. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419850433	6.6	5
536	Update on polyglucosan storage diseases. *Virchows Archiv Fur Pathologische Anatomie Und Physiologie Und Fur Klinische Medizin*, **2019**, 475, 671-686	5.1	13
535	Current and emerging therapies in Becker muscular dystrophy (BMD). *Acta Myologica*, **2019**, 38, 172-179	1.6	8

534	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	
533	AuthorsNreply. *Therapeutic Advances in Neurological Disorders*, **2019**, 12, 1756286419878316	6.6	
532	Clinical and genetic characterization of an Italian family with slow-channel syndrome. *Neurological Sciences*, **2019**, 40, 503-507	3.5	7
531	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
530	MyomiRNAs Dysregulation in ALS Rehabilitation. *Brain Sciences*, **2019**, 9,	3.4	15
529	Microglia polarization by mitochondrial metabolism modulation: A therapeutic opportunity in neurodegenerative diseases. *Mitochondrion*, **2019**, 46, 334-336	4.9	6
528	Heterogeneous Phenotypes in Lipid Storage Myopathy Due to ETFDH Gene Mutations. *JIMD Reports*, **2018**, 38, 33-40	1.9	27
527	A mobile app for patients with Pompe disease and its possible clinical applications. *Neuromuscular Disorders*, **2018**, 28, 471-475	2.9	13
526	Targeted gene panel screening is an effective tool to identify undiagnosed late onset Pompe disease. *Neuromuscular Disorders*, **2018**, 28, 586-591	2.9	14
525	Interpreting Genetic Variants in Titin in Patients With Muscle Disorders. *JAMA Neurology*, **2018**, 75, 557-565	17.2	42
524	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
523	Limb-Girdle Muscular Dystrophy Type 1F **2018**, 41-44		
522	Limb-Girdle Muscular Dystrophy Type 2A **2018**, 45-50		
521	Limb-Girdle Muscular Dystrophy Type 2B **2018**, 51-55		
520	Limb-Girdle Muscular Dystrophy Type 2C **2018**, 57-60		
519	Limb-Girdle Muscular Dystrophy Type 2D **2018**, 61-64		
518	Limb-Girdle Muscular Dystrophy Type 2E **2018**, 65-69		
517	Limb-Girdle Muscular Dystrophy Type 2H **2018**, 73-74		

516 Limb-Girdle Muscular Dystrophy Type 2I **2018**, 75-78

515 Limb-Girdle Muscular Dystrophy Type 2J **2018**, 79-81

514 Duchenne Muscular Dystrophy Carrier **2018**, 9-12

513 Limb-Girdle Muscular Dystrophy Type 2L **2018**, 87-89

512 Limb-Girdle Muscular Dystrophy Type 2T **2018**, 97-98

511 Congenital Muscular Dystrophy Type 1A **2018**, 99-103

510 Fukuyama Congenital Muscular Dystrophy: Walker-Warburg Syndrome **2018**, 107-110

509 Becker Muscular Dystrophy **2018**, 13-16

508 Oculopharyngeal Muscular Dystrophy **2018**, 133-134

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

480	Charcot-Marie-Tooth Neuropathy with Pyramidal Features **2018**, 375-378		
479	Distal Hereditary Motor Neuropathy Type 2C **2018**, 389-390		
478	Limb-Girdle Muscular Dystrophy with Epidermolysis Bullosa Simplex **2018**, 93-95		
477	Myotonic Dystrophy Type 1, Steinert Disease **2018**, 199-203		
476	Duchenne Muscular Dystrophy **2018**, 3-7		1
475	Facioscapulohumeral Muscular Dystrophy Type 1A **2018**, 123-127		
474	Mitochondrial Complex I Deficiency due to ACAD9 Deficiency **2018**, 291-292		1
473	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
472	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
471	An update on diagnostic options and considerations in limb-girdle dystrophies. *Expert Review of Neurotherapeutics*, **2018**, 18, 693-703	4.3	22
470	Enzyme replacement therapy for the treatment of Pompe disease. *Expert Opinion on Orphan Drugs*, **2018**, 6, 311-318	1.1	1
469	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
468	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
467	Copy Number Variants Account for a Tiny Fraction of Undiagnosed Myopathic Patients. *Genes*, **2018**, 9,	4.2	4
466	Regulation of ER-mitochondria contacts by Parkin via Mfn2. *Pharmacological Research*, **2018**, 138, 43-56	10.2	97
465	MicroRNAs are appropriate in mitochondrial related hearing loss? Answer to the skepticism. *Orphanet Journal of Rare Diseases*, **2018**, 13, 119	4.2	1
464	The italian limb girdle muscular dystrophy registry: Relative frequency, clinical features, and differential diagnosis. *Muscle and Nerve*, **2017**, 55, 55-68	3.4	56
463	Elevated Expression of Moesin in Muscular Dystrophies. *American Journal of Pathology*, **2017**, 187, 654-668	3.8	8

462	Micro-RNA expression in muscle and fiber morphometry in myotonic dystrophy type 1. *Neurological Sciences*, **2017**, 38, 619-625	3.5	18
461	The role of transmission electron microscopy in vacuole-associated myopathies. *Ultrastructural Pathology*, **2017**, 41, 88-90	1.3	1
460	Generation of induced Pluripotent Stem Cells as disease modelling of NLSDM. *Molecular Genetics and Metabolism*, **2017**, 121, 28-34	3.7	4
459	Muscle MRI in neutral lipid storage disease (NLSD). *Journal of Neurology*, **2017**, 264, 1334-1342	5.5	11
458	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
457	ATP1A3 mutant patient with alternating hemiplegia of childhood and brain spectroscopic abnormalities. *Journal of the Neurological Sciences*, **2017**, 379, 36-38	3.2	3
456	Cerebral venous thrombosis at high altitude: A systematic review. *Revue Neurologique*, **2017**, 173, 189-193	3	20
455	Limb girdle muscular dystrophies: clinical-genetical diagnostic update and prospects for therapy. *Expert Opinion on Orphan Drugs*, **2017**, 5, 769-784	1.1	6
454	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
453	Targeting deregulated AMPK/mTORC1 pathways improves muscle function in myotonic dystrophy type I. *Journal of Clinical Investigation*, **2017**, 127, 549-563	15.9	45
452	Autophagy dysregulation in Danon disease. *Cell Death and Disease*, **2017**, 8, e2565	9.8	47
451	Aberrant Compartment Formation by HSPB2 Mislocalizes Lamin A and Compromises Nuclear Integrity and Function. *Cell Reports*, **2017**, 20, 2100-2115	10.6	25
450	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
449	Revisiting mitochondrial ocular myopathies: a study from the Italian Network. *Journal of Neurology*, **2017**, 264, 1777-1784	5.5	23
448	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
447	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
446	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
445	Pathogenesis, clinical features and diagnosis of sarcoglycanopathies. *Expert Opinion on Orphan Drugs*, **2016**, 4, 1239-1251	1.1	9

444	Functional changes in Becker muscular dystrophy: implications for clinical trials in dystrophinopathies. *Scientific Reports*, **2016**, 6, 32439	4.9	24
443	The genetic basis of undiagnosed muscular dystrophies and myopathies: Results from 504 patients. *Neurology*, **2016**, 87, 71-6	6.5	70
442	Endocrinological Myopathies **2016**, 143-154		
441	Disease awareness in myotonic dystrophy type 1: an observational cross-sectional study. *Orphanet Journal of Rare Diseases*, **2016**, 11, 34	4.2	45
440	Response to: Mitochondrial neuropathy affects peripheral and cranial nerves and is primary or secondary or both. *Neuromuscular Disorders*, **2016**, 26, 549	2.9	
439	Neuromuscular disease. Diagnosis and discovery in limb-girdle muscular dystrophy. *Nature Reviews Neurology*, **2016**, 12, 6-8	15	4
438	"Mitochondrial neuropathies": A survey from the large cohort of the Italian Network. *Neuromuscular Disorders*, **2016**, 26, 272-6	2.9	32
437	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
436	Circulating microRNAs as biomarkers of muscle differentiation and atrophy in ALS **2016**, 35, 22-30		53
435	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
434	Next generation sequencing detection of late onset pompe disease. *Muscle and Nerve*, **2016**, 53, 981-3	3.4	10
433	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
432	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
431	Challenges and progress in the diagnosis of Congenital Muscular Dystrophies. *Expert Opinion on Orphan Drugs*, **2016**, 4, 347-358	1.1	2
430	Lipolysis and lipophagy in lipid storage myopathies. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2016**, 1862, 1367-73	6.9	22
429	A novel clinical tool to classify facioscapulohumeral muscular dystrophy phenotypes. *Journal of Neurology*, **2016**, 263, 1204-14	5.5	39
428	Progress and challenges in diagnosis of dysferlinopathy. *Muscle and Nerve*, **2016**, 54, 821-835	3.4	38
427	Reply. *Muscle and Nerve*, **2016**, 53, 157-8	3.4	

426	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
425	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
424	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
423	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
422	Clinical and genetic spectrum in limb-girdle muscular dystrophy type 2E. *Neurology*, **2015**, 84, 1772-81	6.5	37
421	Next generation sequencing on patients with LGMD and nonspecific myopathies: Findings associated with ANO5 mutations. *Neuromuscular Disorders*, **2015**, 25, 533-41	2.9	55
420	Redefining phenotypes associated with mitochondrial DNA single deletion. *Journal of Neurology*, **2015**, 262, 1301-9	5.5	53
419	Spectrum of metabolic myopathies. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2015**, 1852, 615-21	6.9	25
418	Dominant muscular dystrophy with a novel SYNE1 gene mutation. *Muscle and Nerve*, **2015**, 51, 145-7	3.4	19
417	TNF- nduced microRNAs Control Dystrophin Expression in Becker Muscular Dystrophy. *Cell Reports*, **2015**, 12, 1678-90	10.6	50
416	Familial polyglucosan body myopathy with unusual phenotype. *Neuropathology and Applied Neurobiology*, **2015**, 41, 385-90	5.2	9
415	Incomplete penetrance in limb-girdle muscular dystrophy type 1F. *Muscle and Nerve*, **2015**, 52, 305-6	3.4	9
414	GYG1 gene mutations in a family with polyglucosan body myopathy. *Neurology: Genetics*, **2015**, 1, e21	3.8	17
413	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
412	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
411	Drugs in development and dietary approach for Duchenne muscular dystrophy. *Orphan Drugs: Research and Reviews*, **2015**, 51		2
410	New Pathogenetic Mechanisms that Link Autophagy to Pompe Disease. *Journal of Neuromuscular Diseases*, **2015**, 2, S9-S9	5	1
409	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

408	Prevalence of asymptomatic vertebral fractures in late-onset Pompe disease. *Journal of Clinical Endocrinology and Metabolism*, **2015**, 100, 401-6	5.6	17
407	Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure. *European Journal of Human Genetics*, **2015**, 23, 1254-8	5.3	36
406	Prevention of cardiomyopathy in Duchenne muscular dystrophy. *Lancet Neurology, The*, **2015**, 14, 127-8	24.1	4
405	Autophagy in Natural History and After ERT in Glycogenosis Type II. *JIMD Reports*, **2015**, 21, 71-7	1.9	4
404	Genetic Modifiers of Duchenne Muscular Dystrophy and Dilated Cardiomyopathy. *PLoS ONE*, **2015**, 10, e0141240	3.7	40
403	Muscle exercise in limb girdle muscular dystrophies: pitfall and advantages. *Acta Myologica*, **2015**, 34, 3-8	1.6	16
402	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
401	Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. *Journal of Neuromuscular Diseases*, **2015**, 2, S36-S37	5	
400	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
399	The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?. *Journal of Neurology*, **2014**, 261, 504-10	5.5	91
398	Skeletal muscle satellite cells in amyotrophic lateral sclerosis. *Ultrastructural Pathology*, **2014**, 38, 295-302	1.3	27
397	Genetic Neuromuscular Disorders **2014**,		4
396	Muscle atrophy, ubiquitin-proteasome, and autophagic pathways in dysferlinopathy. *Muscle and Nerve*, **2014**, 50, 340-7	3.4	27
395	Genotype-phenotype correlation in Pompe disease, a step forward. *Orphanet Journal of Rare Diseases*, **2014**, 9, 102	4.2	44
394	Becker Muscular Dystrophy **2014**, 13-17		
393	G.P.251. *Neuromuscular Disorders*, **2014**, 24, 892	2.9	2
392	Myoclonus in mitochondrial disorders. *Movement Disorders*, **2014**, 29, 722-8	7	27
391	Facioscapulohumeral Muscular Dystrophy Type 1A **2014**, 105-109		

390	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
389	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
388	Duchenne Muscular Dystrophy **2014**, 3-7		
387	"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
386	Muscle fatigue, nNOS and muscle fiber atrophy in limb girdle muscular dystrophy. *Acta Myologica*, **2014**, 33, 119-26	1.6	14
385	Gender difference in limb-girdle muscular dystrophy: a muscle fiber morphometric study in 101 patients **2014**, 33, 179-85		17
384	Duchenne Muscular Dystrophy Carrier **2014**, 9-12		
383	Limb-Girdle Muscular Dystrophy Type 2E **2014**, 61-65		
382	Glycogenosis Type 5, McArdle Disease **2014**, 217-219		
381	Chronic Progressive External Ophthalmoplegia **2014**, 229-232		
380	Congenital Nemaline Myopathy Type 2 **2014**, 137-140		
379	Limb-Girdle Muscular Dystrophy Type 2I **2014**, 71-74		
378	Mitochondrial Encephalomyopathy with COX Deficiency **2014**, 243-246		
377	Limb-Girdle Muscular Dystrophy Type 1C **2014**, 31-34		
376	Myotonic Dystrophy Type 2, Proximal Myotonic Myopathy **2014**, 177-180		
375	Spinal Muscular Atrophy Type 1, Werdnig-Hoffmann Disease **2014**, 295-297		
374	Systemic Primary Carnitine Deficiency **2014**, 261-265		
373	Limb-Girdle Muscular Dystrophy Type 1F **2014**, 35-38		

372	Limb-Girdle Muscular Dystrophy Type 2B **2014**, 47-52		
371	Spinal Muscular Atrophy Type 3, Kugelberg-Welander Disease **2014**, 303-305		
370	Congenital Muscular Dystrophy Type 1A **2014**, 83-87		
369	Glycogenosis Type 2, Pompe Disease **2014**, 205-211		
368	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
367	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
366	Clinical phenotype, muscle MRI and muscle pathology of LGMD1F. *Journal of Neurology*, **2013**, 260, 2033-41	3.5	23
365	Ultrastructural changes in LGMD1F. *Neuropathology*, **2013**, 33, 276-80	2	13
364	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
363	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
362	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
361	Disruption of skeletal muscle mitochondrial network genes and miRNAs in amyotrophic lateral sclerosis. *Neurobiology of Disease*, **2013**, 49, 107-17	7.5	137
360	Phenotypic heterogeneity of the 8344A>G mtDNA "MERRF" mutation. *Neurology*, **2013**, 80, 2049-54	6.5	135
359	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
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357	Recessive MYL2 mutations cause infantile type I muscle fibre disease and cardiomyopathy. *Brain*, **2013**, 136, 282-93	11.2	37
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