Enrico Bertini

List of Publications by Year in Descending Order

701 papers	33,159 citations	91 h-index	148 g-index
759 ext. papers	38,496 ext. citations	5.3 avg, IF	6.57 L-index

#	Paper	IF	Citations
701	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis.. *Neurological Sciences*, **2022**, 43, 2849	3.5	
700	Toward the in vitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development **2022**, 137-150		
699	Safety and efficacy of once-daily risdiplam in type 2 and non-ambulant type 3 spinal muscular atrophy (SUNFISH part 2): a phase 3, double-blind, randomised, placebo-controlled trial.. *Lancet Neurology, The*, **2022**, 21, 42-52	24.1	9
698	Body mass index in type 2 spinal muscular atrophy: a longitudinal study.. *European Journal of Pediatrics*, **2022**, 1	4.1	0
697	Neurological and Neuroimaging Features of -Related Recessive Hereditary Methemoglobinemia Type II.. *Brain Sciences*, **2022**, 12,	3.4	0
696	Therapy Trial Design in Vanishing White Matter: An Expert Consortium Opinion.. *Neurology: Genetics*, **2022**, 8, e657	3.8	1
695	Novel Pathogenic Variants Featuring Unusual Phenotype of Complex Movement Disorder With Thin Corpus Callosum: A Case Report.. *Neurology: Genetics*, **2022**, 8, e661	3.8	
694	Clinical-Genetic Features Influencing Disability in Spastic Paraplegia Type 4: A Cross-sectional Study by the Italian DAISY Network.. *Neurology: Genetics*, **2022**, 8, e664	3.8	0
693	Expanding the clinical-pathological and genetic spectrum of RYR1-related congenital myopathies with cores and minicores: an Italian population study.. *Acta Neuropathologica Communications*, **2022**, 10, 54	7.3	1
692	Genetic modifiers of upper limb function in Duchenne muscular dystrophy.. *Journal of Neurology*, **2022**, 1	5.5	1
691	Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome.. *Molecular Genetics and Metabolism*, **2021**, 135, 109-109	3.7	0
690	Long-term efficacy of T3 analogue Triac in children and adults with MCT8 deficiency: a real-life retrospective cohort study. *Journal of Clinical Endocrinology and Metabolism*, **2021**,	5.6	3
689	Clinical variability at the mild end of BRAT1-related spectrum: Evidence from two families with genotype-phenotype discordance. *Human Mutation*, **2021**,	4.7	1
688	Ataluren delays loss of ambulation and respiratory decline in nonsense mutation Duchenne muscular dystrophy patients. *Journal of Comparative Effectiveness Research*, **2021**,	2.1	6
687	Artificial Intelligence for Dysarthria Assessment in Children With Ataxia: A Hierarchical Approach. *IEEE Access*, **2021**, 9, 166720-166735	3.5	1
686	Clinical, imaging, biochemical and molecular features in Leigh syndrome: a study from the Italian network of mitochondrial diseases. *Orphanet Journal of Rare Diseases*, **2021**, 16, 413	4.2	3
685	haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. *Journal of Medical Genetics*, **2021**,	5.8	3

684	Heterozygous variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. *Journal of Medical Genetics*, **2021**, 58, 475-483	5.8	5
683	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. *Human Mutation*, **2021**, 42, 699-710	4.7	4
682	CASK related disorder: Epilepsy and developmental outcome. *European Journal of Paediatric Neurology*, **2021**, 31, 61-69	3.8	2
681	Refining the mutational spectrum and gene-phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. *Journal of Medical Genetics*, **2021**,	5.8	3
680	Predictive fat mass equations for spinal muscular atrophy type I children: Development and internal validation. *Clinical Nutrition*, **2021**, 40, 1578-1587	5.9	2
679	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. *Disability and Rehabilitation*, **2021**, 1-8	2.4	0
678	Bi-allelic KARS1 pathogenic variants affecting functions of cytosolic and mitochondrial isoforms are associated with a progressive and multisystem disease. *Human Mutation*, **2021**, 42, 745-761	4.7	3
677	Movement Disorders in Children with a Mitochondrial Disease: A Cross-Sectional Survey from the Nationwide Italian Collaborative Network of Mitochondrial Diseases. *Journal of Clinical Medicine*, **2021**, 10,	5.1	3
676	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. *Brain*, **2021**, 144, 3020-3035	11.2	1
675	Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. *Journal of Clinical Medicine*, **2021**, 10,	5.1	3
674	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1622-1634	5.3	6
673	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. *Neuromuscular Disorders*, **2021**, 31, 574-582	2.9	12
672	PIGQ-Related Glycophosphatidylinositol Deficiency Associated with Nonprogressive Congenital Ataxia. *Cerebellum*, **2021**, 1	4.3	1
671	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. *Frontiers in Neurology*, **2021**, 12, 677551	4.1	1
670	North Star Ambulatory Assessment changes in ambulant Duchenne boys amenable to skip exons 44, 45, 51, and 53: A 3 year follow up. *PLoS ONE*, **2021**, 16, e0253882	3.7	1
669	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2021**, 31, 479-488	2.9	
668	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the Mouse Model and Patients. *Frontiers in Physiology*, **2021**, 12, 678974	4.6	0
667	Personalized profiles of antioxidant signaling pathway in patients with tuberculosis. *Journal of Microbiology, Immunology and Infection*, **2021**,	8.5	2

666	Sometimes they come back: New and old spinal muscular atrophy adults in the era of nusinersen. *European Journal of Neurology*, **2021**, 28, 602-608	6	3
665	Clinical phenotypes of infantile onset CACNA1A-related disorder. *European Journal of Paediatric Neurology*, **2021**, 30, 144-154	3.8	4
664	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. *Neurology*, **2021**, 96, e587-e599	6.5	12
663	Novel ACTA1 mutation causes late-presenting nemaline myopathy with unusual dark cores. *Neuromuscular Disorders*, **2021**, 31, 139-148	2.9	1
662	Clinical and radiological profile of patients with spinal muscular atrophy type 4. *European Journal of Neurology*, **2021**, 28, 609-619	6	6
661	Friedreich ataxia in COVID-19 time: current impact and future possibilities. *Cerebellum and Ataxias*, **2021**, 8, 4	1.7	2
660	Type I SMA "new natural history": long-term data in nusinersen-treated patients. *Annals of Clinical and Translational Neurology*, **2021**, 8, 548-557	5.3	10
659	The Spinal Muscular Atrophy Health Index: Italian validation of a disease-specific outcome measure. *Neuromuscular Disorders*, **2021**, 31, 409-418	2.9	2
658	Nuclear Factor Erythroid 2-Related Factor 2 Activation Might Mitigate Clinical Symptoms in Friedreich Ataxia: Clues of an "Out-Brain Origin" of the Disease From a Family Study. *Frontiers in Neuroscience*, **2021**, 15, 638810	5.1	2
657	Mitochondrial Dynamics: Molecular Mechanisms, Related Primary Mitochondrial Disorders and Therapeutic Approaches. *Genes*, **2021**, 12,	4.2	8
656	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. *Neuromuscular Disorders*, **2021**, 31, 596-602	2.9	9
655	Expanded phenotype of AARS1-related white matter disease. *Genetics in Medicine*, **2021**, 23, 2352-2359	8.1	2
654	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. *Muscle and Nerve*, **2021**, 64, 552-559	3.4	4
653	Age-related sensory neuropathy in patients with spinal muscular atrophy type 1. *Muscle and Nerve*, **2021**, 64, 599-603	3.4	
652	Growth patterns in children with spinal muscular atrophy. *Orphanet Journal of Rare Diseases*, **2021**, 16, 375	4.2	3
651	Broadening the spectrum phenotype of TBCE-related neuron neurodegeneration. *Brain and Development*, **2021**, 43, 939-944	2.2	
650	Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases.. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
649	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. *Human Mutation*, **2021**,	4.7	

648	Movement disorders in ADAR1 disease: Insights from a comprehensive cohort. *Parkinsonism and Related Disorders*, **2020**, 79, 100-104	3.6	6
647	Long-term follow-up of patients with type 2 and non-ambulant type 3 spinal muscular atrophy (SMA) treated with olesoxime in the OLEOS trial. *Neuromuscular Disorders*, **2020**, 30, 959-969	2.9	4
646	Oxidative Stress in DNA Repeat Expansion Disorders: A Focus on NRF2 Signaling Involvement. *Biomolecules*, **2020**, 10,	5.9	9
645	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2020**, 30, 492-502	2.9	20
644	Age and sex prevalence estimate of Joubert syndrome in Italy. *Neurology*, **2020**, 94, e797-e801	6.5	11
643	Predictive energy equations for spinal muscular atrophy type I children. *American Journal of Clinical Nutrition*, **2020**, 111, 983-996	7	4
642	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. *Frontiers in Genetics*, **2020**, 11, 131	4.5	25
641	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. *PLoS ONE*, **2020**, 15, e0230677	3.7	15
640	Co-occurrence of mutations in KIF7 and KIAA0556 in Joubert syndrome with ocular coloboma, pituitary malformation and growth hormone deficiency: a case report and literature review. *BMC Pediatrics*, **2020**, 20, 120	2.6	3
639	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. *Frontiers in Genetics*, **2020**, 11, 605	4.5	3
638	The NRF2 Signaling Network Defines Clinical Biomarkers and Therapeutic Opportunity in Friedreich Ataxia. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	17
637	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy Treated with Nusinersen. *Journal of Pediatrics*, **2020**, 219, 223-228.e4	3.6	25
636	TUBB Variants Underlying Different Phenotypes Result in Altered Vesicle Trafficking and Microtubule Dynamics. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	8
635	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. *Annals of Clinical and Translational Neurology*, **2020**, 7, 786-798	5.3	14
634	A wearable video-oculography based evaluation of saccades and respective clinical correlates in patients with early onset ataxia. *Journal of Neuroscience Methods*, **2020**, 338, 108697	3	1
633	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. *Annals of Neurology*, **2020**, 88, 251-263	9.4	21
632	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. *Neurobiology of Disease*, **2020**, 141, 104880	7.5	13
631	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. *Journal of Clinical Investigation*, **2020**, 130, 108-125	15.9	49

630	HDAC inhibitors tune miRNAs in extracellular vesicles of dystrophic muscle-resident mesenchymal cells. *EMBO Reports*, **2020**, 21, e50863	6.5	20
629	Clinical and Genetic Overview of Paroxysmal Movement Disorders and Episodic Ataxias. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	19
628	Speech and Language Disorders in Friedreich Ataxia: Highlights on Phenomenology, Assessment, and Therapy. *Cerebellum*, **2020**, 19, 126-130	4.3	4
627	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. *Clinical Genetics*, **2020**, 97, 521-526	4	7
626	244th ENMC international workshop: Newborn screening in spinal muscular atrophy May 10-12, 2019, Hoofdorp, The Netherlands. *Neuromuscular Disorders*, **2020**, 30, 93-103	2.9	29
625	Systemic activation of Nrf2 pathway in Parkinson disease. *Movement Disorders*, **2020**, 35, 180-184	7	39
624	Genome sequencing in persistently unsolved white matter disorders. *Annals of Clinical and Translational Neurology*, **2020**, 7, 144-152	5.3	13
623	RARS1-related hypomyelinating leukodystrophy: Expanding the spectrum. *Annals of Clinical and Translational Neurology*, **2020**, 7, 83-93	5.3	10
622	Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia. *Computer Methods and Programs in Biomedicine*, **2020**, 188, 105257	6.9	13
621	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. *Neurogenetics*, **2020**, 21, 87-96	3	9
620	SMA THERAPY. *Neuromuscular Disorders*, **2020**, 30, S123-S124	2.9	2
619	PPP1R21-related syndromic intellectual disability: Report of an adult patient and review. *American Journal of Medical Genetics, Part A*, **2020**, 182, 3014-3022	2.5	2
618	Microtubule Dysfunction: A Common Feature of Neurodegenerative Diseases. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	25
617	A Recurrent Pathogenic Variant of Underlies Autosomal Recessive Congenital Muscular Dystrophy With Cataracts and Intellectual Disability: Evidence for a Founder Effect in Southern Italy. *Frontiers in Genetics*, **2020**, 11, 565868	4.5	2
616	Spatio-temporal parameters of ataxia gait dataset obtained with the Kinect. *Data in Brief*, **2020**, 32, 106307	1.2	5
615	Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
614	Validation of low-cost system for gait assessment in children with ataxia. *Computer Methods and Programs in Biomedicine*, **2020**, 196, 105705	6.9	9
613	Aicardi Goutières Syndrome Type 2: A Report on Two Cases with Different Phenotypes Caused by RNASEH2B Gene Mutations. *Journal of Pediatric Neurology*, **2020**, 18, 206-209	0.2	

612	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. *Lancet Diabetes and Endocrinology,the*, **2020**, 8, 594-605	18.1	18
611	Response to "Autosomal recessive axonal neuropathy caused by HINT1 mutation: New association of a psychiatric disorder to the neurological phenotype". *Neuromuscular Disorders*, **2020**, 30, 265-266	2.9	2
610	Ferroptosis in Friedreich Ataxia: A Metal-Induced Neurodegenerative Disease. *Biomolecules*, **2020**, 10,	5.9	9
609	Clinical, neuropathological, and genetic characterization of STUB1 variants in cerebellar ataxias: a frequent cause of predominant cognitive impairment. *Genetics in Medicine*, **2020**, 22, 1851-1862	8.1	16
608	Cardiovascular Involvement in Pediatric Laminopathies. Report of Six Patients and Literature Revision. *Frontiers in Pediatrics*, **2020**, 8, 374	3.4	2
607	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. *Neuromuscular Disorders*, **2020**, 30, 756-764	2.9	13
606	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. *Journal of Comparative Effectiveness Research*, **2020**, 9, 973-984	2.1	19
605	Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. *Oxidative Medicine and Cellular Longevity*, **2020**, 2020, 6821247	6.7	6
604	Clinical Variability in Spinal Muscular Atrophy Type III. *Annals of Neurology*, **2020**, 88, 1109-1117	9.4	14
603	The clinical, histologic, and genotypic spectrum of -related myopathy: A case series. *Neurology*, **2020**, 95, e1512-e1527	6.5	16
602	Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. *Antioxidants*, **2020**, 9,	7.1	2
601	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. *Nephrology Dialysis Transplantation*, **2020**, 35, 1195-1202	4.3	6
600	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients **2020**, 88, 251		1
599	Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. *Orphanet Journal of Rare Diseases*, **2019**, 14, 208	4.2	8
598	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. *American Journal of Human Genetics*, **2019**, 105, 493-508	11	30
597	Nusinersen initiated in infants during the presymptomatic stage of spinal muscular atrophy: Interim efficacy and safety results from the Phase 2 NURTURE study. *Neuromuscular Disorders*, **2019**, 29, 842-856	2.9	188
596	Diagnosis of possible mitochondrial disease: an existential crisis. *Journal of Medical Genetics*, **2019**, 56, 123-130	5.8	27
595	Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. *International Journal of Cardiology*, **2019**, 280, 99-103	3.2	9

594	Clinical, radiological, and genetic characteristics of 16 patients with ACO2 gene defects: Delineation of an emerging neurometabolic syndrome. *Journal of Inherited Metabolic Disease*, **2019**, 42, 264-275	5.4	14
593	Nusinersen in type 1 spinal muscular atrophy: Twelve-month real-world data. *Annals of Neurology*, **2019**, 86, 443-451	9.4	42
592	Long-term natural history data in Duchenne muscular dystrophy ambulant patients with mutations amenable to skip exons 44, 45, 51 and 53. *PLoS ONE*, **2019**, 14, e0218683	3.7	21
591	Molecular Genetics and Interferon Signature in the Italian Aicardi Goutières Syndrome Cohort: Report of 12 New Cases and Literature Review. *Journal of Clinical Medicine*, **2019**, 8,	5.1	16
590	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. *Clinical Genetics*, **2019**, 96, 169-175	4	16
589	Response to Jardim and colleagues regarding comments on "Natural history of a cohort of ABCD1 variant female carriers". *European Journal of Neurology*, **2019**, 26, e77	6	
588	Mutations in ELAC2 associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3′-end processing. *Human Mutation*, **2019**, 40, 1731-1748	4.7	17
587	Mitochondrial Neurodegenerative Disorders II: Ataxia, Dystonia and Leukodystrophies **2019**, 241-256		1
586	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. *PLoS ONE*, **2019**, 14, e0214250	3.7	28
585	An unusual case of late-infantile onset Krabbe disease with selective bilateral corticospinal tract involvement, peripheral demyelinating neuropathy, and mild phenotype. *Acta Neurologica Belgica*, **2019**, 119, 619-620	1.5	0
584	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. *Cerebellum*, **2019**, 18, 422-432	4.3	7
583	Clinical-genetic features and peculiar muscle histopathology in infantile DNM1L-related mitochondrial epileptic encephalopathy. *Human Mutation*, **2019**, 40, 601-618	4.7	22
582	A clinical diagnostic algorithm for early onset cerebellar ataxia. *European Journal of Paediatric Neurology*, **2019**, 23, 692-706	3.8	19
581	Nrf2 Induction Re-establishes a Proper Neuronal Differentiation Program in Friedreich's Ataxia Neural Stem Cells. *Frontiers in Cellular Neuroscience*, **2019**, 13, 356	6.1	21
580	Effectiveness and safety of the tri-iodothyronine analogue Triac in children and adults with MCT8 deficiency: an international, single-arm, open-label, phase 2 trial. *Lancet Diabetes and Endocrinology,the*, **2019**, 7, 695-706	18.1	40
579	Defining the clinical-genetic and neuroradiological features in SPG54: description of eight additional cases and nine novel DDHD2 variants. *Journal of Neurology*, **2019**, 266, 2657-2664	5.5	9
578	Observations from a nationwide vigilance program in medical care for spinal muscular atrophy patients in Chile. *Arquivos De Neuro-Psiquiatria*, **2019**, 77, 470-477	1.6	2
577	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. *Parkinsonism and Related Disorders*, **2019**, 68, 8-16	3.6	13

576	Amish Nemaline Myopathy in 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. *Neuromuscular Disorders*, **2019**, 29, 766-770	2.9	8
575	Evaluation of gait in Duchenne Muscular Dystrophy: Relation of 3D gait analysis to clinical assessment. *Neuromuscular Disorders*, **2019**, 29, 920-929	2.9	4
574	Targeting NRF2 for the Treatment of Friedreich's Ataxia: A Comparison among Drugs. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	32
573	Development of an academic disease registry for spinal muscular atrophy. *Neuromuscular Disorders*, **2019**, 29, 794-799	2.9	17
572	Longitudinal natural history in young boys with Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2019**, 29, 857-862	2.9	12
571	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic pathogenic variants. *Neurology: Genetics*, **2019**, 5, e369	3.8	22
570	Cardiac and Neuromuscular Features of Patients With LMNA-Related Cardiomyopathy. *Annals of Internal Medicine*, **2019**, 171, 458-463	8	22
569	One-year outcome of coenzyme Q10 supplementation in ataxia (ARCA2). *Cerebellum and Ataxias*, **2019**, 6, 15	1.7	7
568	Diagnostic Yield of a Targeted Next-Generation Sequencing Gene Panel for Pediatric-Onset Movement Disorders: A 3-Year Cohort Study. *Frontiers in Genetics*, **2019**, 10, 1026	4.5	12
567	Primary muscle involvement in a 15-year-old girl with the recurrent homozygous c.362dupC variant in FKBP14. *American Journal of Medical Genetics, Part A*, **2019**, 179, 317-321	2.5	1
566	A novel KCTD17 mutation is associated with childhood early-onset hyperkinetic movement disorder. *Parkinsonism and Related Disorders*, **2019**, 61, 4-6	3.6	14
565	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. *Neurological Sciences*, **2019**, 40, 457-468	3.5	16
564	APOPT1/COA8 assists COX assembly and is oppositely regulated by UPS and ROS. *EMBO Molecular Medicine*, **2019**, 11,	12	9
563	SLC2A1 mutations are a rare cause of pediatric-onset hereditary spastic paraplegia. *European Journal of Paediatric Neurology*, **2019**, 23, 329-332	3.8	10
562	Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. *Parkinsonism and Related Disorders*, **2019**, 61, 19-25	3.6	31
561	Natural history of a cohort of ABCD1 variant female carriers. *European Journal of Neurology*, **2019**, 26, 326-332	6	12
560	Neonatal Hypotonia **2019**, 223-233		
559	Novel homozygous GBA2 mutation in a patient with complicated spastic paraplegia. *Clinical Neurology and Neurosurgery*, **2018**, 168, 60-63	2	8

558	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*, **2018**, 28, 103-115	2.9	319
557	Novel Homozygous KCNJ10 Mutation in a Patient with Non-syndromic Early-Onset Cerebellar Ataxia. *Cerebellum*, **2018**, 17, 499-503	4.3	7
556	A V1143F mutation in the neuronal-enriched isoform 2 of the PMCA pump is linked with ataxia. *Neurobiology of Disease*, **2018**, 115, 157-166	7.5	10
555	Clinical and imaging hallmarks of the MYH7-related myopathy with severe axial involvement. *Muscle and Nerve*, **2018**, 58, 224-234	3.4	11
554	A 5-center experience with intrathecal administration of nusinersen in SMA1 in Italy letter to the editor of european journal of pediatric neurology regarding the manuscript "single-center experience with intrathecal administration of nusinersen in children with spinal muscular atrophy type 1" written by pechmann and colleagues". *European Journal of Paediatric Neurology*, **2018**, 22, 729-731	3.8	3
553	ATP1A3-related epileptic encephalopathy responding to ketogenic diet. *Brain and Development*, **2018**, 40, 433-438	2.2	18
552	Serum uric acid in Friedreich Ataxia. *Clinical Biochemistry*, **2018**, 54, 139-141	3.5	6
551	Childhood Rapid-Onset Ataxia: Expanding the Phenotypic Spectrum of ATP1A3 Mutations. *Cerebellum*, **2018**, 17, 489-493	4.3	18
550	Deep intronic variation in splicing regulatory element of the ERCC8 gene associated with severe but long-term survival Cockayne syndrome. *European Journal of Human Genetics*, **2018**, 26, 527-536	5.3	6
549	Neuromyopathy with congenital cataracts and glaucoma: a distinct syndrome caused by POLG variants. *European Journal of Human Genetics*, **2018**, 26, 367-373	5.3	2
548	Clinical, biochemical, and genetic features associated with VARS2-related mitochondrial disease. *Human Mutation*, **2018**, 39, 563-578	4.7	15
547	Expanding the histopathological spectrum of CFL2-related myopathies. *Clinical Genetics*, **2018**, 93, 1234-1239	4	7
546	The impact of next-generation sequencing on the diagnosis of pediatric-onset hereditary spastic paraplegias: new genotype-phenotype correlations for rare HSP-related genes. *Neurogenetics*, **2018**, 19, 111-121	3	28
545	Defective kinesin binding of TUBB2A causes progressive spastic ataxia syndrome resembling sacsinopathy. *Human Molecular Genetics*, **2018**, 27, 1892-1904	5.6	18
544	Compound heterozygous missense and deep intronic variants in NDUFAF6 unraveled by exome sequencing and mRNA analysis. *Journal of Human Genetics*, **2018**, 63, 563-568	4.3	12
543	MRI in sarcoglycanopathies: a large international cohort study. *Journal of Neurology, Neurosurgery and Psychiatry*, **2018**, 89, 72-77	5.5	34
542	Upper limb function in Duchenne muscular dystrophy: 24 month longitudinal data. *PLoS ONE*, **2018**, 13, e0199223	3.7	28
541	Outcome measures for children with mitochondrial disease: consensus recommendations for future studies from a Delphi-based international workshop. *Journal of Inherited Metabolic Disease*, **2018**, 41, 1267-1273	5.4	17

540	An observational study of functional abilities in infants, children, and adults with type 1 SMA. *Neurology*, **2018**, 91, e696-e703	6.5	12
539	Nusinersen in type 1 SMA infants, children and young adults: Preliminary results on motor function. *Neuromuscular Disorders*, **2018**, 28, 582-585	2.9	53
538	Nonprogressive congenital ataxias. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2018**, 155, 91-103	3	19
537	X-linked ataxias. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2018**, 155, 175-189	3	10
536	Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. *Neuromuscular Disorders*, **2018**, 28, 197-207	2.9	236
535	A randomized placebo-controlled phase 3 trial of an antisense oligonucleotide, drisapersen, in Duchenne muscular dystrophy. *Neuromuscular Disorders*, **2018**, 28, 4-15	2.9	68
534	Longitudinal gait assessment in a stiff person syndrome. *International Journal of Rehabilitation Research*, **2018**, 41, 377-379	1.8	5
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532	Non-invasive Focal Mechanical Vibrations Delivered by Wearable Devices: An Open-Label Pilot Study in Childhood Ataxia. *Frontiers in Neurology*, **2018**, 9, 849	4.1	9
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