Enrico Bertini

List of Publications by Year
in descending order

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727 papers	42,990 citations	2565 99 h-index	6024 165 g-index
759 all docs	759 docs citations	759 times ranked	38800 citing authors

#	Article	IF	Citations
1	Refining the mutational spectrum and geneâ€“phenotype correlates in pontocerebellar hypoplasia: results of a multicentric study. Journal of Medical Genetics, 2022, 59, 399-409.	1.5	13
2	PIGQ-Related Glycophosphatidylinositol Deficiency Associated with Nonprogressive Congenital Ataxia. Cerebellum, 2022, 21, 525-530.	1.4	2
3	Personalized profiles of antioxidant signaling pathway in patients with tuberculosis. Journal of Microbiology, Immunology and Infection, 2022, 55, 405-412.	1.5	3
4	SUFU haploinsufficiency causes a recognisable neurodevelopmental phenotype at the mild end of the Joubert syndrome spectrum. Journal of Medical Genetics, 2022, 59, 888-894.	1.5	19
5	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, 2022, 107, e1136-e1147.	1.8	15
6	Revised upper limb module in type II and III spinal muscular atrophy: 24-month changes. Neuromuscular Disorders, 2022, 32, 36-42.	0.3	13
7	Clinical variability at the mild end of BRAT1 â€related spectrum: Evidence from two families with genotypeâ€“phenotype discordance. Human Mutation, 2022, 43, 67-73.	1.1	9
8	Ataluren delays loss of ambulation and respiratory decline in nonsense mutation Duchenne muscular dystrophy patients. Journal of Comparative Effectiveness Research, 2022, 11, 139-155.	0.6	29
9	A case of spastic paraplegia type 11 mimicking a GM2-gangliosidosis. Neurological Sciences, 2022, 43, 2849-2852.	0.9	0
10	Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley Syndrome. Molecular Genetics and Metabolism, 2022, 135, 109-113.	0.5	17
11	Toward the inÂvitro understanding of iPSC nucleoskeletal and cytoskeletal biology, and their relevance for organoid development. , 2022, , 137-150.		0
12	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.	4.9	89
13	Body mass index in type 2 spinal muscular atrophy: a longitudinal study. European Journal of Pediatrics, 2022, , 1.	1.3	2
14	Cerebellar Agenesis and Bilateral Polimicrogyria Associated with Rare Variants of CUB and Sushi Multiple Domains 1 Gene (CSMD1): A Longitudinal Neuropsychological and Neuroradiological Case Study. International Journal of Environmental Research and Public Health, 2022, 19, 1224.	1.2	0
15	Neurological and Neuroimaging Features of CYB5R3-Related Recessive Hereditary Methemoglobinemia Type II. Brain Sciences, 2022, 12, 182.	1.1	4
16	Therapy Trial Design in Vanishing White Matter. Neurology: Genetics, 2022, 8, e657.	0.9	12
17	Upper Body Physical Rehabilitation for Children with Ataxia through IMU-Based Exergame. Journal of Clinical Medicine, 2022, 11, 1065.	1.0	7
18	Nusinersen efficacy data for 24â€month in type 2 and 3 spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2022, 9, 404-409.	1.7	22

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19	Modeling PCDH19-CE: From 2D Stem Cell Model to 3D Brain Organoids. International Journal of Molecular Sciences, 2022, 23, 3506.	1.8	1
20	Novel SEPSECS Pathogenic Variants Featuring Unusual Phenotype of Complex Movement Disorder With Thin Corpus Callosum. Neurology: Genetics, 2022, 8, e661.	0.9	0
21	Clinical-Genetic Features Influencing Disability in Spastic Paraplegia Type 4. Neurology: Genetics, 2022, 8, e664.	0.9	9
22	Clinical and Genetic Aspects of Phelanâ€“McDermid Syndrome: An Interdisciplinary Approach to Management. Genes, 2022, 13, 504.	1.0	9
23	A novel homozygous variant in <scp>COX5A</scp> causes an attenuated phenotype with failure to thrive, lactic acidosis, hypoglycemia, and short stature. Clinical Genetics, 2022, 102, 56-60.	1.0	3
24	Response to: Phenotypic heterogeneity of Leigh syndrome due to NDUFA12 variants is multicausal. Human Mutation, 2022, 43, 99-100.	1.1	0
25	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.	2.4	3
26	Genetic modifiers of upper limb function in Duchenne muscular dystrophy. Journal of Neurology, 2022, 269, 4884-4894.	1.8	2
27	â€œAtypicalâ€•Krabbe disease in two siblings harboring biallelic GALC mutations including a deep intronic variant. European Journal of Human Genetics, 2022, , .	1.4	4
28	Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome. Brain, 2022, 145, 3383-3390.	3.7	3
29	Superior Cerebellar Atrophy: An Imaging Clue to Diagnose ITPR1-Related Disorders. International Journal of Molecular Sciences, 2022, 23, 6723.	1.8	4
30	New Insights into the Neurodegeneration Mechanisms Underlying Riboflavin Transporter Deficiency (RTD): Involvement of Energy Dysmetabolism and Cytoskeletal Derangement. Biomedicines, 2022, 10, 1329.	1.4	5
31	Heterozygous KIF1A variants underlie a wide spectrum of neurodevelopmental and neurodegenerative disorders. Journal of Medical Genetics, 2021, 58, 475-483.	1.5	21
32	Sometimes they come back: New and old spinal muscular atrophy adults in the era of nusinersen. European Journal of Neurology, 2021, 28, 602-608.	1.7	9
33	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	0.7	13
34	Respiratory Trajectories in Type 2 and 3 Spinal Muscular Atrophy in the iSMAC Cohort Study. Neurology, 2021, 96, e587-e599.	1.5	36
35	Novel ACTA1 mutation causes late-presenting nemaline myopathy with unusual dark cores. Neuromuscular Disorders, 2021, 31, 139-148.	0.3	4
36	Clinical and radiological profile of patients with spinal muscular atrophy type 4. European Journal of Neurology, 2021, 28, 609-619.	1.7	23

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37	Friedreich ataxia in COVID-19 time: current impact and future possibilities. Cerebellum and Ataxias, 2021, 8, 4.	1.9	8
38	Type I SMA â€œnew natural historyâ€• longâ€term data in nusinersenâ€treated patients. Annals of Clinical and Translational Neurology, 2021, 8, 548-557.	1.7	35
39	The Spinal Muscular Atrophy Health Index: Italian validation of a disease-specific outcome measure. Neuromuscular Disorders, 2021, 31, 409-418.	0.3	7
40	Nuclear Factor Erythroid 2-Related Factor 2 Activation Might Mitigate Clinical Symptoms in Friedreichâ€™s Ataxia: Clues of an â€œOut-Brain Originâ€•of the Disease From a Family Study. Frontiers in Neuroscience, 2021, 15, 638810.	1.4	5
41	Altered cytoskeletal arrangement in induced pluripotent stem cells and motor neurons from patients with riboflavin transporter deficiency. DMM Disease Models and Mechanisms, 2021, 14, .	1.2	5
42	Mitochondrial Dynamics: Molecular Mechanisms, Related Primary Mitochondrial Disorders and Therapeutic Approaches. Genes, 2021, 12, 247.	1.0	25
43	Novel NDUFA12 variants are associated with isolated complex I defect and variable clinical manifestation. Human Mutation, 2021, 42, 699-710.	1.1	12
44	CASK related disorder: Epilepsy and developmental outcome. European Journal of Paediatric Neurology, 2021, 31, 61-69.	0.7	7
45	Predictive fat mass equations for spinal muscular atrophy type I children: Development and internal validation. Clinical Nutrition, 2021, 40, 1578-1587.	2.3	3
46	Challenges and resources in adult life with Joubert syndrome: issues from an international classification of functioning (ICF) perspective. Disability and Rehabilitation, 2021, , 1-8.	0.9	1
47	Biâ€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.	1.1	7
48	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, 2063.	1.0	8
49	Biallelic mutations in RNF220 cause laminopathies featuring leukodystrophy, ataxia and deafness. Brain, 2021, 144, 3020-3035.	3.7	11
50	Dissecting the Role of PCDH19 in Clustering Epilepsy by Exploiting Patient-Specific Models of Neurogenesis. Journal of Clinical Medicine, 2021, 10, 2754.	1.0	13
51	Nusinersen in pediatric and adult patients with type III spinal muscular atrophy. Annals of Clinical and Translational Neurology, 2021, 8, 1622-1634.	1.7	27
52	Newborn screening programs for spinal muscular atrophy worldwide: Where we stand and where to go. Neuromuscular Disorders, 2021, 31, 574-582.	0.3	94
53	The ARCA Registry: A Collaborative Global Platform for Advancing Trial Readiness in Autosomal Recessive Cerebellar Ataxias. Frontiers in Neurology, 2021, 12, 677551.	1.1	15
54	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.	1.1	6

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55	The nonsense mutation stop+4 model correlates with motor changes in Duchenne muscular dystrophy. Neuromuscular Disorders, 2021, 31, 479-488.	0.3	0
56	Circadian Genes as Exploratory Biomarkers in DMD: Results From Both the mdx Mouse Model and Patients. Frontiers in Physiology, 2021, 12, 678974.	1.3	1
57	Age related treatment effect in type II Spinal Muscular Atrophy pediatric patients treated with nusinersen. Neuromuscular Disorders, 2021, 31, 596-602.	0.3	29
58	Expanded phenotype of AARS1-related white matter disease. Genetics in Medicine, 2021, 23, 2352-2359.	1.1	8
59	Different trajectories in upper limb and gross motor function in spinal muscular atrophy. Muscle and Nerve, 2021, 64, 552-559.	1.0	18
60	Ageâ€related sensory neuropathy in patients with spinal muscular atrophy type 1. Muscle and Nerve, 2021, 64, 599-603.	1.0	3
61	Growth patterns in children with spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2021, 16, 375.	1.2	19
62	SMA-miRs (miR-181a-5p, -324-5p, and -451a) are overexpressed in spinal muscular atrophy skeletal muscle and serum samples. ELife, 2021, 10, .	2.8	13
63	Broadening the spectrum phenotype of TBCE-related neuron neurodegeneration. Brain and Development, 2021, 43, 939-944.	0.6	0
64	Expanding the Clinical and Mutational Spectrum of the PLP1-Related Hypomyelination of Early Myelinated Structures (HEMS). Brain Sciences, 2021, 11, 93.	1.1	3
65	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.	1.2	16
66	Myocardial and Arrhythmic Spectrum of Neuromuscular Disorders in Children. Biomolecules, 2021, 11, 1578.	1.8	5
67	Artificial Intelligence for Dysarthria Assessment in Children With Ataxia: A Hierarchical Approach. IEEE Access, 2021, 9, 166720-166735.	2.6	4
68	Induced Pluripotent Stem Cells (iPSCs) and Gene Therapy: A New Era for the Treatment of Neurological Diseases. International Journal of Molecular Sciences, 2021, 22, 13674.	1.8	13
69	Impaired urinary concentration ability is a sensitive predictor of renal disease progression in Joubert syndrome. Nephrology Dialysis Transplantation, 2020, 35, 1195-1202.	0.4	15
70	Speech and Language Disorders in Friedreich Ataxia: Highlights on Phenomenology, Assessment, and Therapy. Cerebellum, 2020, 19, 126-130.	1.4	12
71	Hereditary spastic paraplegia is a novel phenotype for germline de novo ATP1A1 mutation. Clinical Genetics, 2020, 97, 521-526.	1.0	14
72	244th ENMC international workshop: Newborn screening in spinal muscular atrophy May 10â€“12, 2019, Hoofdorp, The Netherlands. Neuromuscular Disorders, 2020, 30, 93-103.	0.3	55

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73	Systemic Activation of Nrf2 Pathway in Parkinson's Disease. Movement Disorders, 2020, 35, 180-184.	2.2	66
74	Genome sequencing in persistently unsolved white matter disorders. Annals of Clinical and Translational Neurology, 2020, 7, 144-152.	1.7	26
75	RARS1â€related hypomyelinating leukodystrophy: Expanding the spectrum. Annals of Clinical and Translational Neurology, 2020, 7, 83-93.	1.7	18
76	Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia. Computer Methods and Programs in Biomedicine, 2020, 188, 105257.	2.6	21
77	Mitochondrial epilepsy: a cross-sectional nationwide Italian survey. Neurogenetics, 2020, 21, 87-96.	0.7	14
78	SMA â€“ THERAPY. Neuromuscular Disorders, 2020, 30, S123-S124.	0.3	6
79	<scp>PPP1R21</scp>â€related syndromic intellectual disability: Report of an adult patient and review. American Journal of Medical Genetics, Part A, 2020, 182, 3014-3022.	0.7	8
80	Long-Term Outcome of LVAD in Duchenne Population with End Stage Cardiomyopathy. Journal of Heart and Lung Transplantation, 2020, 39, S219-S220.	0.3	0
81	Microtubule Dysfunction: A Common Feature of Neurodegenerative Diseases. International Journal of Molecular Sciences, 2020, 21, 7354.	1.8	63
82	A Recurrent Pathogenic Variant of INPP5K 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.	1.1	8
83	Spatio-temporal parameters of ataxia gait dataset obtained with the Kinect. Data in Brief, 2020, 32, 106307.	0.5	10
84	Antioxidant Amelioration of Riboflavin Transporter Deficiency in Motoneurons Derived from Patient-Specific Induced Pluripotent Stem Cells. International Journal of Molecular Sciences, 2020, 21, 7402.	1.8	8
85	Validation of low-cost system for gait assessment in children with ataxia. Computer Methods and Programs in Biomedicine, 2020, 196, 105705.	2.6	17
86	Aicardiâ€“GoutiÃ¨res Syndrome Type 2: A Report on Two Cases with Different Phenotypes Caused by RNASEH2B Gene Mutations. Journal of Pediatric Neurology, 2020, 18, 206-209.	0.0	0
87	Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes and Endocrinology,the, 2020, 8, 594-605.	5.5	50
88	Response to â€œAutosomal recessive axonal neuropathy caused by HINT1 mutation: New association of a psychiatric disorder to the neurological phenotypeâ€• Neuromuscular Disorders, 2020, 30, 265-266.	0.3	4
89	Ferroptosis in Friedreichâ€™s Ataxia: A Metal-Induced Neurodegenerative Disease. Biomolecules, 2020, 10, 1551.	1.8	21
90	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.	1.1	30

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91	Cardiovascular Involvement in Pediatric Laminopathies. Report of Six Patients and Literature Revision. Frontiers in Pediatrics, 2020, 8, 374.	0.9	9
92	Age and baseline values predict 12 and 24-month functional changes in type 2 SMA. Neuromuscular Disorders, 2020, 30, 756-764.	0.3	25
93	Meta-analyses of ataluren randomized controlled trials in nonsense mutation Duchenne muscular dystrophy. Journal of Comparative Effectiveness Research, 2020, 9, 973-984.	0.6	41
94	Mitochondrial and Peroxisomal Alterations Contribute to Energy Dysmetabolism in Riboflavin Transporter Deficiency. Oxidative Medicine and Cellular Longevity, 2020, 2020, 1-19.	1.9	13
95	Clinical Variability in Spinal Muscular Atrophy Type <scp>III</scp>. Annals of Neurology, 2020, 88, 1109-1117.	2.8	34
96	The clinical, histologic, and genotypic spectrum of SEPN1 -related myopathy. Neurology, 2020, 95, e1512-e1527.	1.5	44
97	Mitochondrial Abnormalities in Induced Pluripotent Stem Cells-Derived Motor Neurons from Patients with Riboflavin Transporter Deficiency. Antioxidants, 2020, 9, 1252.	2.2	11
98	Movement disorders in ADAR1 disease: Insights from a comprehensive cohort. Parkinsonism and Related Disorders, 2020, 79, 100-104.	1.1	6
99	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.	0.3	15
100	Oxidative Stress in DNA Repeat Expansion Disorders: A Focus on NRF2 Signaling Involvement. Biomolecules, 2020, 10, 702.	1.8	17
101	Randomized phase 2 trial and open-label extension of domagrozumab in Duchenne muscular dystrophy. Neuromuscular Disorders, 2020, 30, 492-502.	0.3	40
102	Age and sex prevalence estimate of Joubert syndrome in Italy. Neurology, 2020, 94, e797-e801.	1.5	26
103	Predictive energy equations for spinal muscular atrophy type I children. American Journal of Clinical Nutrition, 2020, 111, 983-996.	2.2	8
104	The Genetic Landscape of Dystrophin Mutations in Italy: A Nationwide Study. Frontiers in Genetics, 2020, 11, 131.	1.1	49
105	Diagnostic journey in Spinal Muscular Atrophy: Is it still an odyssey?. PLoS ONE, 2020, 15, e0230677.	1.1	38
106	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.	0.7	12
107	Tumor Necrosis Factor Receptor SF10A (TNFRSF10A) SNPs Correlate With Corticosteroid Response in Duchenne Muscular Dystrophy. Frontiers in Genetics, 2020, 11, 605.	1.1	9
108	The NRF2 Signaling Network Defines Clinical Biomarkers and Therapeutic Opportunity in Friedreichâ€™s Ataxia. International Journal of Molecular Sciences, 2020, 21, 916.	1.8	27

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109	Respiratory Needs in Patients with Type 1 Spinal Muscular Atrophy TreatedÂwith Nusinersen. Journal of Pediatrics, 2020, 219, 223-228.e4.	0.9	51
110	TUBB Variants Underlying Different Phenotypes Result in Altered Vesicle Trafficking and Microtubule Dynamics. International Journal of Molecular Sciences, 2020, 21, 1385.	1.8	20
111	Genetic modifiers of respiratory function in Duchenne muscular dystrophy. Annals of Clinical and Translational Neurology, 2020, 7, 786-798.	1.7	36
112	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.	1.3	3
113	Clinicoâ€Genetic, Imaging and Molecular Delineation of <scp>COQ8A</scp>â€Ataxia: A Multicenter Study of 59 Patients. Annals of Neurology, 2020, 88, 251-263.	2.8	52
114	A homozygous MRPL24 mutation causes a complex movement disorder and affects the mitoribosome assembly. Neurobiology of Disease, 2020, 141, 104880.	2.1	29
115	Clinico-Genetic, Imaging and Molecular Delineation of COQ8A-Ataxia: A Multicenter Study of 59 Patients. , 2020, 88, 251.		1
116	HDAC inhibitors tune miRNAs in extracellular vesicles of dystrophic muscleâ€resident mesenchymal cells. EMBO Reports, 2020, 21, e50863.	2.0	45
117	Clinical and Genetic Overview of Paroxysmal Movement Disorders and Episodic Ataxias. International Journal of Molecular Sciences, 2020, 21, 3603.	1.8	36
118	A clinical diagnostic algorithm for early onset cerebellar ataxia. European Journal of Paediatric Neurology, 2019, 23, 692-706.	0.7	37
119	Nrf2 Induction Re-establishes a Proper Neuronal Differentiation Program in Friedreichâ€™s Ataxia Neural Stem Cells. Frontiers in Cellular Neuroscience, 2019, 13, 356.	1.8	36
120	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.	5.5	77
121	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.	1.8	19
122	Observations from a nationwide vigilance program in medical care for spinal muscular atrophy patients in Chile. Arquivos De Neuro-Psiquiatria, 2019, 77, 470-477.	0.3	6
123	Dystonia-Ataxia with early handwriting deterioration in COQ8A mutation carriers: A case series and literature review. Parkinsonism and Related Disorders, 2019, 68, 8-16.	1.1	25
124	â€˜Amish Nemaline Myopathyâ€™ in 2 Italian siblings harbouring a novel homozygous mutation in Troponin-I gene. Neuromuscular Disorders, 2019, 29, 766-770.	0.3	13
125	Evaluation of gait in Duchenne Muscular Dystrophy: Relation of 3D gait analysis to clinical assessment. Neuromuscular Disorders, 2019, 29, 920-929.	0.3	14
126	Targeting NRF2 for the Treatment of Friedreichâ€™s Ataxia: A Comparison among Drugs. International Journal of Molecular Sciences, 2019, 20, 5211.	1.8	45

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127	Development of an academic disease registry for spinal muscular atrophy. Neuromuscular Disorders, 2019, 29, 794-799.	0.3	29
128	Longitudinal natural history in young boys with Duchenne muscular dystrophy. Neuromuscular Disorders, 2019, 29, 857-862.	0.3	23
129	Corticospinal tract damage in HHH syndrome: a metabolic cause of hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2019, 14, 208.	1.2	12
130	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.	2.6	48
131	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.	0.3	401
132	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	1.5	42
133	Heart rate reduction strategy using ivabradine in end-stage Duchenne cardiomyopathy. International Journal of Cardiology, 2019, 280, 99-103.	0.8	17
134	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.	1.7	18
135	Nusinersen in type 1 spinal muscular atrophy: Twelveâ€month realâ€world data. Annals of Neurology, 2019, 86, 443-451.	2.8	83
136	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.	1.1	47
137	Molecular Genetics and Interferon Signature in the Italian Aicardi GoutiÃ¨res Syndrome Cohort: Report of 12 New Cases and Literature Review. Journal of Clinical Medicine, 2019, 8, 750.	1.0	29
138	Heterozygous missense variants of SPTBN2 are a frequent cause of congenital cerebellar ataxia. Clinical Genetics, 2019, 96, 169-175.	1.0	27
139	Response to Jardim and colleagues regarding comments on â€˜Natural history of a cohort of <scp>ABCD</scp>1 variant female carriersâ€™. European Journal of Neurology, 2019, 26, e77.	1.7	0
140	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	1.1	31
141	Mitochondrial Neurodegenerative Disorders II: Ataxia, Dystonia and Leukodystrophies. , 2019, , 241-256.		1
142	Targeting ferroptosis: A novel therapeutic strategy for the treatment of mitochondrial disease-related epilepsy. PLoS ONE, 2019, 14, e0214250.	1.1	59
143	An unusual case of late-infantile onset Krabbe disease with selective bilateralÂcorticospinal tract involvement, peripheral demyelinating neuropathy, and mild phenotype. Acta Neurologica Belgica, 2019, 119, 619-620.	0.5	1
144	Biallelic Variants in the Nuclear Pore Complex Protein NUP93 Are Associated with Non-progressive Congenital Ataxia. Cerebellum, 2019, 18, 422-432.	1.4	10

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145	Clinical-genetic features and peculiar muscle histopathology in infantileDNM1L-related mitochondrial epileptic encephalopathy. Human Mutation, 2019, 40, 601-618.	1.1	31
146	Cardiac and Neuromuscular Features of Patients WithLMNA-Related Cardiomyopathy. Annals of Internal Medicine, 2019, 171, 458.	2.0	33
147	One-year outcome of coenzyme Q10 supplementation in ADCK3 ataxia (ARCA2). Cerebellum and Ataxias, 2019, 6, 15.	1.9	15
148	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.	1.1	33
149	Primary muscle involvement in a 15â€yearâ€old girl with the recurrent homozygous c.362dupC variant in FKBP14. American Journal of Medical Genetics, Part A, 2019, 179, 317-321.	0.7	3
150	A novel KCTD17 mutation is associated with childhood early-onset hyperkinetic movement disorder. Parkinsonism and Related Disorders, 2019, 61, 4-6.	1.1	22
151	Italian recommendations for diagnosis and management of congenital myasthenic syndromes. Neurological Sciences, 2019, 40, 457-468.	0.9	24
152	APOPT 1/ COA 8 assists COX assembly and is oppositely regulated by UPS and ROS. EMBO Molecular Medicine, 2019, 11, .	3.3	19
153	SLC2A1 mutations are a rare cause of pediatric-onset hereditary spastic paraplegia. European Journal of Paediatric Neurology, 2019, 23, 329-332.	0.7	11
154	Phenomenology and clinical course of movement disorder in GNAO1 variants: Results from an analytical review. Parkinsonism and Related Disorders, 2019, 61, 19-25.	1.1	64
155	Natural history of a cohort of <scp>ABCD</scp>1 variant female carriers. European Journal of Neurology, 2019, 26, 326-332.	1.7	19
156	Neonatal Hypotonia. , 2019, , 223-233.		0
157	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	3.9	65
158	Clinical spectrum of POLR3-related leukodystrophy caused by biallelic POLR1C pathogenic variants. Neurology: Genetics, 2019, 5, e369.	0.9	38
159	Novel homozygous GBA2 mutation in a patient with complicated spastic paraplegia. Clinical Neurology and Neurosurgery, 2018, 168, 60-63.	0.6	9
160	Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders, 2018, 28, 103-115.	0.3	584
161	Novel Homozygous KCNJ10 Mutation in a Patient with Non-syndromic Early-Onset Cerebellar Ataxia. Cerebellum, 2018, 17, 499-503.	1.4	10
162	A V1143F mutation in the neuronal-enriched isoform 2 of the PMCA pump is linked with ataxia. Neurobiology of Disease, 2018, 115, 157-166.	2.1	15

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163	Clinical and imaging hallmarks of the MYH7â€related myopathy with severe axial involvement. Muscle and Nerve, 2018, 58, 224-234.	1.0	14
164	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 â€œsingle-center experience with intrathecal administration of nusinersen in children with spinal muscular atrophy type 1â€• written by pechmann and colleaguesâ€• European Journal of Paediatric Neurology, 2018, 22, 729-731.	0.7	5
165	ATP1A3 -related epileptic encephalopathy responding to ketogenic diet. Brain and Development, 2018, 40, 433-438.	0.6	23
166	Serum uric acid in Friedreich Ataxia. Clinical Biochemistry, 2018, 54, 139-141.	0.8	7
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